

Exhibit 84

**UNITED STATES DISTRICT COURT
DISTRICT OF NEW JERSEY**

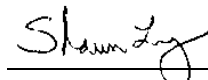
**IN RE JOHNSON & JOHNSON
TALCUM POWDER PRODUCTS
MARKETING, SALES PRACTICES, AND
PRODUCTS LIABILITY LITIGATION**

THIS DOCUMENT RELATES TO ALL CASES

MDL NO. 16-2738 (MAS) (RLS)

**AMENDED RULE 26 EXPERT REPORT OF
SHAWN LEVY, PHD**

Dated: November 15, 2023



Shawn Levy, PhD

The following amended report is provided pursuant to Rule 26 of the Federal Rules of Civil Procedure. My opinions are as follows:

I. Qualifications and Background

I am the Chief Scientific Officer at Element Biosciences and formerly the Chief Scientific Officer of Discovery Life Sciences. In my current role, I focus on developing disruptive DNA sequencing technology on the AVITI platform. My work is centered around applications, methodology and product strategy for use in research and the commercial market, as well as research partnerships and collaboration with academia, industry and commercial entities. This work includes performance assessment for DNA sequencing and developing techniques for testing and diagnosing patients with undiagnosed and rare diseases. I also have ongoing topical research with collaborations in various areas such as changes in immune cell gene expression with environmental changes, genetic risk and sex differences in brain disorders, use of AI in genome interpretation, transcriptome changes in COVID-19 patients, alternative splicing in betacoronaviruses, and transcriptome analyses during immune surveillance and cell proliferation.

I was the founding director of the Genomic Services Laboratory and a faculty investigator at the HudsonAlpha Institute for Biotechnology. I previously served as executive director of the HudsonAlpha Clinical Services Laboratory, LLC, which I launched in 2014. I am adjunct faculty in the Department of Genetics and Department of Epidemiology at the University of Alabama at Birmingham, adjunct faculty in the Department of Biological Sciences at the University of Alabama at Huntsville. Prior to joining the faculty at HudsonAlpha, I was a faculty member in the Department of Molecular Physiology and Biophysics and the Department of Biomedical Informatics at Vanderbilt University Medical Center from 2000 until 2009. I began at the rank of Research Assistant Professor and advanced to Assistant Professor prior to joining HudsonAlpha in 2009. I continued my appointment at Vanderbilt as an Adjunct Associate Professor of Biomedical Informatics after joining HudsonAlpha's faculty.

My academic positions were continuously funded, primarily from the NIH, for nearly 20 years. My extramural funding support has been in excess of \$100M and I have been a principal investigator in programs such as the Gabriella Miller Kids First Program and the All of Us

Research Program. I have been an author or co-author on over 200 peer-reviewed publications that have over 55,000 citations. I serve as an ad hoc reviewer for scientific journals, including Nature, Nature Genetics, Science, Cell, Genome Research, and several others. I have been a co-chair of the Genomics Working Group of the American Medical Informatics Association, a community of scientists and healthcare professionals that work to facilitate collaboration and share knowledge across a continuum, from basic and applied research to the consumer and public health arenas.

I received my Ph.D. in biochemistry and completed a postdoctoral fellowship in genetics at Emory University in Atlanta, where I set up a microarray facility at the Emory Center for Molecular Medicine. My education, training, and experience are further set forth in my Curriculum Vitae (CV), attached to this report as **Exhibit A**.

As detailed in my CV, my research activities have examined several fundamental questions in human cancer, such as the role of viral infection in head and neck cancer, the role of genetic mutation in risk for secondary cancer events following initial treatment, the genetics of B-cell lymphoma, hepatosplenic T-cell lymphoma and malignant melanoma, and the role of STAT3 in triple-negative breast cancer. As the founding and Executive Director of the HudsonAlpha Clinical Services Laboratory, I engaged in research interests and responsibilities in the clinical use of genetic testing for cancer risk and treatment stratification. HudsonAlpha launched the Information is Power campaign and provided genetic testing for breast and ovarian cancer risk to women across Alabama free of charge. My lab at HudsonAlpha supported the Alabama Genomics Health Initiative, which tests for genetic risks and carrier status for several diseases, including breast and ovarian cancer. This body of work in basic and clinical research, in combination with earlier epidemiological work in the Shanghai Women's Health study, provides the experience, education, and expertise to develop this report.

I have been retained to describe the role of genetics in the pathogenesis of cancer in general and specifically ovarian cancer. I have been asked to assess whether perineal use of talcum powder products induces a biologically plausible mechanism or mechanisms that result in ovarian cancer. Lastly, I have been asked to evaluate the genetic testing results of six individual plaintiffs and to opine on the impact, if any, the results had on their development of ovarian cancer.

My report consists of a review of existing works and materials from various sources, including primary literature, review articles, and other cited sources, and my conclusions regarding

this cause-and-effect relationship. My opinions are based on assessing and weighing the totality of the evidence, including relevant literature and available documentation, and my experience as a geneticist and scientific researcher. The methodology I have used to reach my opinions in this case is generally accepted in the scientific community and is the same methodology I use in my research and other professional activities.

My opinions below are held to a reasonable degree of scientific certainty. My opinions reflect my sole and independent judgment at the time of this report.

A list of the materials I have considered is attached to this report as **Exhibit B**. My billing rate is \$500 per hour. I have not testified by deposition or at trial during the last four years.

II. Cancer Overview

Cancer has become a descriptor that is ubiquitously used but describes an extraordinarily complex and diverse collection of medical conditions. Cancer is also a word representing an amazingly complicated and often misunderstood collection of diseases. Cancer is a disease of unregulated cell growth at the most basic level. Cancer's simplicities end with that brief description. From conception until death, humans experience an unending cycle of cell growth, differentiation, and death. As infants grow to children and then to adults, there is an array of growth processes that occur that represent the milestones of development and maturation, many of which continue throughout adulthood. These processes are an orchestra of highly coordinated and regulated events with essential checks and balances. When those highly regulated processes are defective or the checks and balances malfunction, the growth of the cells can become unregulated. Which tissue or cells become unregulated, and precisely what process is impacted defines the type of cancer and its progression. Some cancer types can be aggressive and highly metastatic when unregulated cells invade other body parts and destroy organs and tissues. Other types of cancer remain restricted to specific organs or cell types and may be less aggressive.

The DNA within our cells provides the genetic code or instructions to create the cells, tissues, and organs that make humans. Subtle changes in that code lead to the diversity of people worldwide, while more substantial changes in that code create the diversity of life forms around us, from the smallest bacteria to the largest plants and animals. All cells have one set of instructions that provides the information for cells to divide, tissues to grow, and how cells should die.

III. The Role of Gene Mutations in the Development of Cancer

Cancer is a general term used to describe conditions of unregulated cell growth that disrupt normal cellular and organ processes. Changes in cell growth lead to a wide variety of disease phenotypes that make up the diversity of cancers. Cancers are typically dictated by the tissue origin of the unregulated growth and the extent of metastasis to other tissues or organs. Cancer is a disease caused by DNA changes (mutations) that disrupt normal cell growth and regulation. Our DNA is organized into thousands of individual genes, each containing a specific subset of instructions telling the cell what functions to perform and how to grow and divide. Mutations that cause cancer most commonly disrupt the regulation of the cell cycle (i.e., stages of cell growth and division). The following classifications of mutations are commonly found in cancer, but many other gene mutations can also contribute.

Increasing cell growth and division. A gene mutation can initiate rapid cell growth and division, resulting in many new cells with the same mutation. Proto-oncogenes are a group of genes that regulate cell growth, differentiation, division, and death. When a proto-oncogene is mutated, it can become an oncogene that then instructs the cell to grow rapidly in an unregulated manner.

Loss of growth inhibition. A gene mutation can result in the renewed growth of a previously quiescent cell. Normal cells regulate their division so that the human body contains the appropriate number of each cell type. When the tumor suppressor genes that provide this inhibitory control become mutated, cells become cancer cells and continue to grow and amass. An example of one such gene is *TP53*, which is discussed in more detail below.

Loss of DNA repair. Gene mutations can also affect the genes that proofread DNA and fix mutations before they can have a detrimental effect. DNA repair genes detect errors in cellular DNA and correct them. A mutation in a DNA repair gene may mean that other errors aren't corrected, leading cells to become cancerous through unchecked replication of damaged cells. Examples of DNA repair genes include *BRCA1* and *BRCA2*, which are discussed in more detail below.

Gene mutations can be classified by when they occur.

- 1) Inherited gene mutations: Inherited gene mutations are those mutations an individual is born with and is present in all body cells. These types of mutations define traits and characteristics that have a family history. This type of mutation directly accounts for a small percentage of cancers. The indirect effects of this type of mutation are an area of active research. A growing number of genes and mutations are known to increase the risk of cancer. Specific mutations in *BRCA1* and *BRCA2* increase the risk for breast and ovarian cancer by disrupting DNA repair, for example. While additional gene mutations associated with cancer are being identified, the percentage of individuals affected by those mutations will be significantly less than those affected by *BRCA1* and *BRCA2*.
- 2) Acquired (somatic) gene mutations: Somatic mutations are acquired after birth. Most gene mutations that directly cause cancer occur after birth and aren't inherited. Several events or exposures can cause gene mutations. These include environmental exposures such as smoking, radiation, and cancer-causing chemicals (carcinogens). Biological and lifestyle exposures such as viruses, hormones, and chronic inflammation are also known to result in cancer-causing mutations. Each exposure type has its own mechanism for increasing the risk of cancer. These mechanisms may be direct, such as radiation directly damaging DNA or asbestos causing mesothelioma, or indirect, such as an external agent causing a cellular reaction or inflammatory response that leads to DNA damage or mutation.

Both inherited and acquired gene mutations work together to cause cancer. While genetic testing has become commonplace for assessing risk for cancer and directing treatment, the catalog of oncogenes, tumor suppressor genes, and DNA repair genes make genetic testing valuable and impactful for informing patients of their genetic risk for cancer. Genetic testing generally detects inherited mutations. Genetic screening does not detect acquired gene mutations because they occur only in specific cells. Inheriting a genetic mutation that increases risk to cancer doesn't guarantee that cancer will occur. One or more additional mutations may be needed to initiate or cause cancer, and the inherited mutation could increase the chance of metastasis or disease progression. The inherited gene mutation could increase susceptibility to cancer when someone is exposed to a specific cancer-causing substance. For example, chemical and other environmental agents, such as talcum powder products, can cause mutations that, acting along with inherited mutations, cause

ovarian cancer. Conversely, individuals may still develop cancer if they do not have mutations known to predispose them to cancer.

In summary, cancer does not begin and progress in a straightforward manner. Instead, it occurs on a spectrum. Transitioning from normal to metastatic disease is not a simple on/off switch. It involves numerous steps, pauses, and evolutions that vary between individuals and cancer types.

IV. The Role of Genetics in Ovarian Cancer

Ovarian cancer is the primary cause of death from gynecologic disease and the second most common gynecologic malignancy in the United States.¹ Since the pathogenesis, treatment, and clinical courses are similar, the term "ovarian cancer" often includes ovarian epithelial cancer, fallopian tube cancer, and peritoneal cancers. Researchers now believe most of these cancers originate in the distal portion of the fallopian tube (Levanon, Crum and Drapkin 2008). The significant mortality is primarily associated with late diagnosis and resistance to therapy (Bowtell 2010). Epithelial ovarian cancer (EOC) includes most malignant ovarian neoplasms (Chan, Cheung et al. 2006) that can be classified based on morphologic and molecular genetic features into the following types: serous (OSC; low and high grade), endometrioid (EC), clear cell (OCCC) and mucinous (MC) carcinomas.

Certain specific genetic and transcriptional signatures are associated with each histological subtype. Low-grade OSC cases generally have genetic alterations in *BRAF*, *KRAS*, *NRAS*, and Erb-B2 Receptor Tyrosine Kinase 2 (*ERBB2*); high-grade OSC has mutations in Tumor Protein P53 (*TP53*), *BRCA1/2*, Neurofibromin 1 (*NF1*), RB Transcriptional Corepressor 1 (*RBI*), and Cyclin-Dependent Kinase 12 (*CDK12*) (Chan, Cheung et al. 2006). Homologous recombination repair of DNA damage is defective in approximately 50% of high-grade serous cancers along with alterations in signaling pathways such as PI3, Ras, Notch, and FoxM1 (Nunes and Serpa 2018).

Endometrioid carcinoma (EC) subtypes are associated with mutations in AT-Rich Interaction Domain 1A (*ARID1A*), Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha (*PI3KCA*), Phosphatase and Tensin Homolog (*PTEN*), Protein Phosphatase 2

¹ <https://www.cdc.gov/cancer/ovarian/statistics/index.htm>

Scaffold Subunit Alpha (*PPP2R1α*), and mismatch repair deficiency. Ovarian clear cell carcinoma (OCCC) subtypes have been found with de novo expression of *HNF1β* (Mabuchi, Kawase et al. 2009, Shen, Fridley et al. 2013) as well as inherited mutations in *ARID1A*, *PI3KCA*, *PTEN*, Catenin Beta 1 (*CTNNB1*) and *PPP2R1α*. MC comprises tumors associated with inherited mutations in *KRAS* and a high frequency of *ERBB2* amplification with overexpression of mucin-coding genes (Banerjee and Kaye 2013, Jayson, Kohn et al. 2014).

In addition to inherited mutations, environmental exposure can result in DNA changes or acquired gene mutations that lead to cancer. These sources can be from exposure to minerals such as asbestos, talc, or nickel, chemical exposures such as benzene or formaldehyde, and natural radiation sources like radon or ultraviolet light. These exposures constantly damage human DNA. Fortunately, cells have robust DNA repair mechanisms to ensure DNA damage is repaired before the DNA is replicated. These “proofreading” mechanisms react to DNA damage and stop DNA replication. The mechanisms involve checkpoint control proteins such as the p53 protein, which halts the cell cycle if DNA is damaged, thus suppressing tumor production. Cells that do not express functional p53 protein exhibit high mutation rates in response to DNA damage, accelerating the formation of tumors.

BRCA1 and BRCA2 proteins also function in the DNA repair pathway. *BRCA1* and *BRCA2* are expressed in breast and other tissue cells and help repair damaged DNA, or destroy cells if DNA cannot be repaired. They are involved in the repair of chromosomal damage resulting from double-strand breaks. BRCA1 functions as part of a protein complex called BRCA1-associated genome surveillance complex (BASC). BASC combines with several other proteins to form a large complex that senses DNA damage. BRCA2 is a complex protein that, among other things, interacts with the RAD51 protein, creating a complex vital for DNA repair (Andreassen, Seo et al. 2021).

Individuals can inherit *BRCA1*, *BRCA2*, or *TP53*² mutations and are termed “positive” for the gene mutation. Such mutations will detrimentally affect the ability to repair DNA or sense the presence of damaged DNA. These defects allow additional mutations to accumulate in cells, leading to a higher probability of cells becoming cancerous. *BRCA1*, *BRCA2*, and *TP53* mutations

² Genes consist of genetic information that code for functional proteins. Both the gene and the protein they code share the same alphanumeric name. To avoid confusion, genes are italicized in text and proteins are not. For example: *BRCA1* (gene) and BRCA1 (protein).

can also be acquired in specific cells. If those cells form a tumor, the cancerous tissue can be tested for these gene mutations.

BRCA mutations are inherited in an autosomal dominant fashion, meaning inheriting only one copy (a monoallelic, or heterozygous mutation) results in increased cancer risk. Some individuals with a heterozygous mutation in the *BRCA1* or *BRCA2* gene will develop cancer during their lifetime, but others will not. Penetrance refers to the proportion of individuals with a genetic mutation who exhibit symptoms of the disorder. Where some carriers do not develop a disorder, as in the case of *BRCA* carriers, the condition is said to have incomplete penetrance. In such instances, additional genetic, environmental, and lifestyle factors must be present for the disorder to manifest. The lifetime risk for ovarian cancer is approximately 40 percent for *BRCA1* carriers and 15 to 20 percent for *BRCA2* carriers (Berek, Crum and Friedlander 2012, Paluch-Shimon, Cardoso et al. 2016). Therefore, the presence of mutations in the *BRCA* genes does not guarantee that carriers will get cancer. These mutations make a person more susceptible to developing cancer when exposed to a carcinogen (Park, 2018; Vitonis, 2011; Wu, 2015).

Mutations in *BRCA* genes are found in the minority of epithelial ovarian cancer cases, suggesting additional mechanisms involving other genes that predispose women to ovarian cancer. The location of the mutation within the *BRCA1* and *BRCA2* genes has been associated with different ovarian cancer risk (Thompson, Valdimarsdottir et al. 2002, Coppa, Buffone et al. 2014, Bayraktar, Jackson et al. 2015). Additionally, several common alleles, or alternate forms of a gene, have been found to modify ovarian cancer risk for *BRCA1* and *BRCA2* mutation carriers. These modifier genes alter how information from a gene is used to synthesize a final gene product (gene expression), which in turn causes a disease. They are hypothesized to act as low to moderate penetrance alleles contributing to ovarian cancer risk. (Ramus, Vierkant et al. 2008, Sellers, Huang et al. 2008, Barnes and Antoniou 2012, Saed, Diamond and Fletcher 2017). These modifiers consist of changes in the DNA called single-nucleotide variants (SNVs), resulting in a point mutation in the gene. If the mutation causes a change in protein amino acid, the mutation can result in a functionally or structurally altered protein. If the mutation does not change an amino acid or the amino acid change does not alter function or structure, the mutation may be benign. Some affected proteins are oxidants, antioxidants, or otherwise involved in regulatory pathways involving cancer risk, as discussed below.

Lynch syndrome is another hereditary condition that increases the risk of ovarian cancer. It is caused by mutations that impair DNA mismatch repair, and the disease is inherited in an autosomal dominant manner similar to *BRCA* mutations. As in the case of *BRCA* mutations, due to incomplete penetrance, inheriting a Lynch-associated mutation does not guarantee an individual will get cancer. Still, the risk of cancer will increase when exposed to a carcinogen.

Myriad Genetics was an early pioneer in developing commercial genetic testing for *BRCA1* and *BRCA2* mutations and predicting breast and ovarian cancer risk. As with all inherited traits, a positive family history is the strongest indicator of genetic risk alleles in an individual. Since the exact identity of those risk alleles and the magnitude of cancer risk remain unknown until testing is performed, early guidelines for testing were based on a positive family history. The availability of testing has increased, and the costs of testing have fallen. However, genetic testing remains a relatively rare practice in the general population. Since the early 1990s, advanced molecular biological technologies have allowed for the connection to be made between specific genetic mutations and the resulting hereditary cancers. The genes involved in cancer development are well established because of the large number of individuals tested and the ability to trace their genetic inheritance. The overall spectrum has additional variants and genes with minor involvement. For example, genetic testing can identify variants of unknown significance (VUS), which have insufficient or conflicting evidence associating them with a disease. More discussion of genetic testing and variants of unknown significance is included in section VII below. The implications, if any, of those variants are difficult to discern due to their rarity, and the likelihood that their action depends upon additional specific and complex interactions that occur in rare situations involving a combination of variations and dysregulation. The history of *BRCA1* and *BRCA2* testing serves as an important example of the complexities of cancer genetics and the challenges that still remain. This story is well summarized in a recent publication by Toland and colleagues from the Breast Cancer Information Core (BIC) Steering Committee (Toland, Brody and Committee 2019). The last decade of genetic analysis has revealed extensive genetic variation in cancer, making discerning highly penetrant driver mutations from variable passenger mutations a statistical analysis (Forbes, Beare et al. 2015, Wishart 2015). The fact that few genes have been identified with significant effect size indicates that it is unlikely that additional genes with effects sizes like *BRCA1* and 2 have been missed in breast and ovarian cancer. Therefore, it is improbable

that any emerging variants could have a single-gene effect size like known mutations such as *BRCA1* or *BRCA2*.

V. Response to Cellular Injury

As mentioned, the human body requires continuous cell growth and development for normal everyday health and function. Some tissues and cell types continually turn over. Our skin, blood cells, immune cells, and the cells that line our digestive tract are examples of tissues or cells that are constantly growing and replacing older or damaged cells. In the case of an injury, a complex cascade of events begins, which involves inflammation and culminates in wound healing. During tissue injury, cell proliferation is enhanced while the tissue regenerates. After the healing is complete, proliferation and inflammation subside.

If proliferating cells sustain DNA damage or mutations that alter cell growth or cell regulation, those cells may continue to proliferate in certain microenvironments. Those environments are often rich in inflammatory cells and growth factors that support cell growth. Essentially, this is like a positive feedback cycle of injury and attempted regeneration and growth. Recent studies have shown a link between inflammation associated with wound healing and ovarian cancer cell seeding (Jia, Nagaoka et al. 2018). In addition to inflammation, the innate immune response plays a role in promoting cancer development and progression. These observations are generally accepted in the scientific literature (Coussens and Werb 2002, Pardoll 2002).

VI. Inflammation

A. The Role of Inflammation in Cancer - General

The functional relationship between cancer and inflammation was first described in the mid-1800s. Rudolf Virchow noted leucocytes in neoplastic tissues in 1863 and made a connection between inflammation and cancer (as cited in Balkwill and Mantovani, 2001). He noted that the "lymphoreticular infiltrate" was reflective of the cancer origin at sites of chronic inflammation. Research published over the last 20 years has provided a further understanding of the inflammatory microenvironment of malignant tissues and validates Virchow's hypothesis. Furthermore, the links between cancer and inflammation now have solid implications for prevention and treatment.

Macrophages are versatile immune-system cells that play various roles in health and well-being. They act in tissues and free-floating cells in the blood that engulf and digest cellular debris, foreign substances, infectious microbes, cancer cells, and anything that does not have the correct cell surface proteins to indicate a healthy cell to the body. They take various forms with various names throughout the body and have specialized tasks, including recruiting other immune cells like lymphocytes to sites of infection or acting as antigen-presenting cells to T cells. Upon activation by contact with substances foreign to the body, macrophages release small proteins called cytokines. Generally speaking, macrophages can increase or decrease inflammation depending on the cytokines released.

Tumor-associated macrophages (TAM) are a significant component of the infiltrate of most, if not all, tumors (Franklin and Li 2016). TAM derive from circulating monocytic precursors and are directed into the tumor by chemoattractant cytokines called chemokines. Tumor cells often prolong the survival of TAM by producing cytokines called colony-stimulating factors. When appropriately activated, TAM can kill tumor cells or elicit tissue destructive reactions on the vascular endothelium to disrupt blood supply to the tumor. However, TAM also produces growth and angiogenic factors and protease enzymes that degrade the extracellular matrix. Therefore, TAM can stimulate tumor-cell proliferation, promote angiogenesis, and favor invasion and metastasis (Mantovani, Bussolino and Dejana 1992, Mantovani, Bussolino and Introna 1997). Direct evidence for the importance of protease production by TAM, neutrophils, and mast cells during experimental carcinogenesis was reported more than 15 years ago (Coussens, Tinkle et al. 2000). Since that time, the report by Coussens has been cited nearly 300 times by other studies. This dual potential of TAM has been described in the literature as the "macrophage balance." (Mantovani, Bottazzi et al. 1992, Liu and Cao 2015).

B. The Role of Inflammation in Ovarian Cancer

Inflammation has also been shown to play a vital role in epithelial ovarian cancer. This principle is generally accepted in the scientific community and very well reviewed in the scientific literature over the last fifteen years, as the role of inflammation is typical in many types of cancer. (Pardoll 2002, Shan and Liu 2009, Mor, Yin et al. 2011, Pejovic and Nezhat 2011, Maccio and Madeddu 2012, Charbonneau, Goode et al. 2013, Kisielewski, Tolwinska et al. 2013). The literature reviews and many direct studies feature the immune system as an essential mediator of

ovarian carcinogenesis via two models for its role in ovarian cancer: 1) chronic inflammation and 2) incessant ovulation.

- 1) Chronic Inflammation: The chronic inflammation model of carcinogenesis proposes that chronic exposures to external or endogenous triggers of immunity (such as known carcinogens) and the persistence of immune cells cause ovarian cancer. These inflammatory triggers cause injury to surrounding epithelium, damage DNA by releasing reactive oxygen species (ROS), or produce cytokines that promote proliferation (Saed, Diamond and Fletcher 2017). One environmental exposure that induces inflammation in animal models and human lungs is talcum powder (Wehner 1994). Composed primarily of magnesium silicate (though it also contains asbestos, heavy metals, and other inflammatory agents as described below), the genital use of talcum powder has been linked to ovarian cancer risk in several studies (Ness, Grisso et al. 2000, Mills, Riordan et al. 2004, Merritt, Green et al. 2008, Wu, Pearce et al. 2009, Rosenblatt, Weiss et al. 2011, Penninkilampi and Eslick 2018).
- 2) Incessant Ovulation: As stated in (Charbonneau, Goode et al. 2013), incessant ovulation results in damage due to rupturing of the ovulating follicle, which traumatizes the ovarian surface, causing an immediate inflammatory response and wound repair. Repeating this process of damage and epithelial proliferation to repair the wound increases the risk of malignant transformation. Epidemiologic studies beginning nearly 50 years ago have implicated an increased number of ovulations as a risk factor for ovarian cancer (Mahdavi, Pejovic and Nezhat 2006). In contrast, decreased risk of (i.e., protection from) ovarian cancer has been associated with increased parity (Adami, Hsieh et al. 1994, Modan, Hartge et al. 2001), oral contraceptive use (Narod, Risch et al. 1998), breastfeeding (Jordan, Cushing-Haugen et al. 2012) and older age at first menses (Titus-Ernstoff, Perez et al. 2001). All of these protective factors impact the number of lifetime ovulations. One of these early studies from the late 1970s, which more recent investigations have further substantiated, found protective effects of “anovulatory time” by combining information on both increased oral contraceptive use and parity as well as age at first and last menses (Casagrande, Louie et al. 1979), supporting the theory of incessant ovulation as an underlying mechanism of carcinogenesis.

As a part of the inflammatory response, macrophages induce oxidative stress by producing reactive oxygen species (ROS) and reactive nitrogen species (RNS). Typically, oxidants and antioxidants maintain a balance wherein the amount of ROS does not overwhelm the body's and antioxidants' ability to regulate them. Free radicals such as ROS and RNS are highly reactive and adversely alter DNA, proteins, and lipids (which comprise cell membranes) to promote tumor development and progression. Many cancers arise from sites with chronic irritation, infection, or inflammation. Cancer cells persist in a pro-oxidant state with excess production and ROS generation that allows for tumor initiation, promotion, and progression.

The association between exposure to pathogens and chronic inflammation in tumor promotion and progression further supports the generally understood principle that chronic inflammation plays a crucial role in the development of ovarian cancer. Examples of inflammatory conditions associated with ovarian cancer include endometriosis and pelvic inflammatory disease. Evidence strongly suggests that endometriosis is a pelvic inflammatory condition (Agic, Xu et al. 2006), and that inflammation explains the association between endometriosis and epithelial ovarian cancer (Ness, Grisso et al. 2000). Studies have found a relationship between pelvic inflammatory disease and ovarian cancer risk (Merritt, Green et al. 2008, Lin, Tu et al. 2011). Moreover, the effect of non-steroidal anti-inflammatory drugs (NSAIDs) to reduce the risk of ovarian cancer provides additional support. The earlier studies focusing on NSAIDs were preliminary and results were somewhat inconsistent (Bonovas, Filioussi and Sitaras 2005, Merritt, Green et al. 2008). Still, a more recent pooled analysis examining 12 case-control studies found aspirin could reduce ovarian cancer risk by 20%-34% (Trabert, Ness et al. 2014). Moreover, the protective effect of aspirin is maintained even in individuals with genetic susceptibility to ovarian cancer (Hurwitz, Webb et al. 2023).

Additional studies illustrate the potential protective effects of anti-inflammatory agents, including unexpected drugs such as metformin. As reviewed in (Reid, Permuth and Sellers 2017), evidence supports a role of the anti-diabetic agent, metformin, in preventing and treating multiple cancers (Li 2011). Studies reviewed include a case-control study including 1,611 incident ovarian cancer cases performed using the UK-based General Practice Research Database (Bodmer, Becker et al. 2011). Long-term use (≥ 30 prescriptions) of metformin (and not sulfonylureas or insulin) was associated with a trend towards reduced risk with an odds ratio of 0.61. Though these results

alone were not statistically significant, the observation that the anti-inflammatory agent, metformin, appears to decrease cancer risk is evidence that inflammation is a primary mediator of ovarian cancer. (Irie, Banno et al. 2016).

Considering the well-established role of inflammation in cancer and the beneficial effects of anti-inflammatory compounds on cancer risk and progression, it is logical to examine the environmental factors that may directly lead to cancer or increased chronic inflammation and indirectly lead to cancer. The International Agency for Research on Cancer (IARC) has recognized for nearly thirty years that there is sufficient evidence to conclude human exposure to asbestos is a cause of ovarian cancer (IARC 1987, IARC 1987, IARC 2012). Not surprisingly, human studies have reported asbestos fibers in ovaries (Heller, Gordon et al. 1996, Langseth, Johansen et al. 2007). Meta-analysis supports the conclusion that exposure to asbestos increases the risk of ovarian cancer (Camargo, Stayner et al. 2011).

C. Talcum Powder Products

Numerous studies have examined the role of talcum powder use in developing ovarian cancers. A comprehensive and recent meta-analysis by Penninkilampi found an association between perineal talc use and ovarian cancer, with a more significant association after a higher number of lifetime applications (Penninkilampi and Eslick 2018). The Penninkilampi study identified 24 case-control (13,421 cases) and three cohort studies (890 cases). Observational studies involving 50 cases or more of ovarian cancer were deemed eligible for inclusion. Penninkilampi analyzed the association between ovarian cancer and any perineal talc use. Included studies reported specific types of ovarian cancer, long-term (>ten years) talc use, total lifetime applications, frequency, and use of talc while also using diaphragms or sanitary napkins.

The Penninkilampi study found a consistent association between perineal talc use and ovarian cancer. Variation in the magnitude of the effect was found when considering the study design and ovarian cancer subtype. Any perineal talc use was associated with an increased risk of ovarian cancer (OR=1.31, 95%CI 1.24-1.39). Greater than 3,600-lifetime applications (OR=1.42, 95%CI 1.25-1.61) were slightly more associated with ovarian cancer than less than 3,600 applications (OR=1.32, 95%CI 1.15- 1.50). Another meta-analysis by Taher, et al. in 2019, reviewing the same studies as Penninkilampi, found a similar increased risk of ovarian cancer with perineal use of talcum powder (Kadry Taher, Farhat et al. 2019). A more recent meta-analysis

focusing on frequent use (at least twice per week), concluded the increased risk of ovarian cancer with perineal exposure was 31-65% (Woolen, Lazar and Smith-Bindman 2022).

In addition to epidemiological evidence, an *in vitro* experiment by Buz'Zard and Lau reported an increase in ROS generation, increased cell proliferation, and neoplastic transformation (conversion into cancerous cells) in human ovarian cells treated with talcum powder (Buz'Zard and Lau 2007). They also found talcum powder treatment increased the number of reactive oxygen species produced by polymorphonuclear neutrophils, inflammatory cells whose role is to release significant quantities of reactive oxygen species in response to various harmful foreign stimuli. Additional studies have also shown the effects of talc on the immune response (Hamilton, Fox et al. 1984, National Toxicology 1993, Keskin, Teksen et al. 2009).

Some studies have suggested that a link between ovarian cancer and talcum powder product use may be influenced by several genes (Gates, Tworoger et al. 2008, Shukla, MacPherson et al. 2009, Fletcher, Harper et al. 2019). Gates and colleagues found that women with specific genetic variants in glutathione S-transferase M1 (*GSTM1*) and/or glutathione S-transferase T1 (*GSTT1*) may have a higher risk of ovarian cancer associated with talc use (Gates, Tworoger et al. 2008).

In another study, talcum powder increased mRNA levels of pro-oxidant enzymes in normal ovarian epithelial cells and ovarian cancer cell lines while decreasing the mRNA levels of antioxidant enzymes (Fletcher, Harper et al. 2019). Talcum powder exposure to normal and epithelial ovarian cancer cells resulted in an increased pro-oxidant state as evidenced by observing increased levels of CA-125, caspase-3, nitrate/nitrite, and key redox enzymes (Fletcher, Harper et al. 2019). CA-125 is significant because it is a biomarker that is elevated in patients with ovarian cancer and is currently FDA-approved for disease monitoring in patients with epithelial ovarian cancer, as well as those with BRCA mutations or in another high-risk group. Mandarino and colleagues independently published separate work showing the pro-oxidant effect of talc in a cell culture system. The same effects were not observed with titanium dioxide or airborne particulates (Mandarino, Gregory et al. 2020). In the same study Mandarino also reported an immune modulating effect of talc to exposed macrophages. Supporting observations of the immune-modulating effect of talc were made by Emi and colleagues in a separate study (Emi, Rivera et al. 2021).

Multiple epidemiological studies examining aggregate data from large cohorts of patients have consistently found an increased risk of talc use and ovarian cancer (Penninkilampi and Eslick

2018, Wentzensen and O'Brien 2021). Two independent research studies examined cohorts of women with self-reported use of talc, store-bought douches, or a combination of both (Gabriel, Vitonis et al. 2019, O'Brien, D'Aloisio et al. 2019). The two studies found a positive association between talc use with or without douching and ovarian cancer. Still, neither found an association of douching alone with the risk for ovarian cancer. A separate research study examined several pro-inflammatory stimuli to develop an inflammation-related risk score (IRRS) and determine the association of that risk score to ovarian cancer progression and survival (Brieger, Phung et al. 2022). As reported by Brieger et al., the study used pooled data from 8,147 women with invasive epithelial ovarian cancer from the Ovarian Cancer Association Consortium. Pre-diagnosis inflammatory-related exposures included alcohol use, aspirin use, other nonsteroidal anti-inflammatory drug use, body mass index, environmental tobacco smoke exposure, history of pelvic inflammatory disease, polycystic ovarian syndrome, endometriosis, menopausal hormone therapy use, physical inactivity, smoking status, and talc use. The study's results indicated a statistically significant trend of increasing risk of death per quartile of the IRRS (HR=1.09, 95% CI 1.03–1.14). The study reported that women in the upper quartile of the IRRS had a 31% higher death rate than the lowest (95% CI 1.11–1.54) (Brieger, Phung et al. 2022). Although the work by Brieger and colleagues does not determine the role of any single activity or exposure in disease progression or survival, it adds additional evidence supporting the role of inflammation in the development, progression, and survival of ovarian cancer. Inflammation from any source is potentially harmful, and when inflammation is introduced chronically to the genital region using external compounds such as talcum powder, the risk of developing ovarian cancer increases. The Brieger study demonstrates that increased inflammation is associated with poor survival in advanced ovarian cancer.

D. Constituents of Talcum Powder Products - Asbestos, Fibrous Talc, Heavy Metals and Fragrance Chemicals

In addition to the platy talc, I have seen evidence that talcum powder products, including Johnson's Baby Powder and Shower to Shower, contain asbestos³ and heavy metals⁴, such as

³ Ex. 28 and Ex. D-1A, Hopkins Dep. (Aug. 16 & 17, 2018; Oct. 26, 2018; and Nov. 5, 2018); Blount, 1991; Paoletti, 1984.

⁴ Ex. 47, Julie Pier Dep. (Sept. 12 & 13, 2018).

chromium, cobalt, and nickel. A 2017 study by Longo and Rigler on historic samples of Johnson & Johnson Baby Powder ranging in production date over many years showed over one-half (17 of 30) of the samples contained asbestos (Longo and Rigler 2017). Fibrous talc was found in 15 of the 30 samples. A 2018 study by Longo and Rigler reported both the presence of fibrous anthophyllite and fibrous talc in products tested from 1978 (Longo and Rigler 2018). Additionally, I have reviewed the expert report of Drs. Longo and Rigler reporting that 37 of 56 historical talcum powder samples contained amphibole asbestos and 41 of the 42 samples tested contained fibrous talc.⁵ In 2019, the FDA tested Johnson's Baby Powder and found that it contained chrysotile asbestos and talc fibers.⁶

Asbestos has long been recognized as a carcinogen, and exposure can cause mesothelioma, and cancers of the lung, larynx, and ovary (IARC 1987, IARC 2012). It is established that asbestos exposure can result in macrophage activation, inflammation, generation of reactive oxygen and reactive nitrogen species, tissue injury, genotoxicity, and resistance to programmed cell death (IARC 2012). One of the direct mechanisms is through interactions between internalized fibers and components of mitosis, resulting in chromosomal alterations and abnormalities (Hesterberg, Butterick et al. 1986, Wang, Jaurand et al. 1987, Yegles, Saint-Etienne et al. 1993). IARC has classified asbestos as a known human carcinogen (Group 1). Human tumors resulting from asbestos exposure can be characterized by genetic and chromosomal alterations that lead to the inactivation of tumor-suppressor genes (IARC 2012).

Talc not containing asbestiform fibers has been found by IARC to be a Group 2b or "possible" carcinogen (IARC, 2010). IARC has determined that fibrous talc or talc-containing asbestiform fibers (talc occurring in a fibrous habit) is a carcinogen to humans (IARC 2012).

IARC classifies chromium and nickel as Group 1, "carcinogenic to humans" (IARC 2012). Cobalt is classified as Group 2B, "possibly carcinogenic to humans" (IARC 2006). IARC defines possibly carcinogenic as "a positive association has been observed between exposure to the agent and cancer for which a causal interpretation has been considered by the Working Group to be credible, but chance, bias or confounding could not be ruled out with reasonable confidence." Established mechanisms of chromium carcinogenesis include DNA damage, mutation, genomic

⁵ Amended Expert Report of William E. Longo, PhD and Mark W. Rigler, PhD (February 2, 2019) (finding that 68% of the samples tested contained amphibole asbestos and 98% contained fibrous talc or talc fibers).

⁶ AMA Analytical Services, Inc. – Certificate of Analysis – Job Name: Task 3 – Analysis of Official Samples; Job Number: CLIN 1 – Task 3 (Oct. 11, 2019).

instability, and cell transformation (Straif, Benbrahim-Tallaa et al. 2009). Similar mechanisms result from nickel exposure (IARC, 2012). Cobalt exposure has been shown to cause increased production of reactive oxygen species and other inflammatory and proliferative changes (IARC, 2006).

I also reviewed Dr. Michael Crowley's report discussing the numerous fragrance chemicals added to talcum powder products. I agree with Dr. Crowley's opinion that these chemicals contribute to the inflammatory properties, toxicity, and potential carcinogenicity of the products. The presence of these constituents as part of talcum powder products provides additional evidence of biological plausibility for talcum powder and ovarian cancer.⁷

Carcinogenesis is a complex and dynamic process that occurs due to a combination of genetic and acquired mutations in an individual along with other processes. Mutations arising from environmental sources have an additive and possibly multiplicative effect toward ultimately causing carcinogenesis (Vitonis, Titus-Ernstoff and Cramer 2011, Wu, Pearce et al. 2015, Park, Schildkraut et al. 2018). The presence of asbestos, fibrous talc, nickel, and chromium, known carcinogens, in talcum powder products further supports the conclusion that talcum powder causes chronic inflammation.

Based on these observations and lines of evidence, it is my opinion that talcum powder causes inflammation, which initiates a biological response that includes oxidative stress, cell proliferation, inhibition of apoptosis, and genetic mutations, which result in cancer development and progression. This process explains the biologically plausible mechanism for talcum powder products causing ovarian cancer.

VII. Genetic Testing in Oncology

A. Background

The principles of genetics or how we inherit traits and features from generation to generation have been studied for hundreds of years. Major advancements in understanding the mechanisms of genetics and role that DNA plays in health and disease have advanced rapidly over the last several decades. From discovering the structure of DNA to sequencing the first human genome to now being able to routinely sequence the genome of any individual, the advancements

⁷ Expert Report of Michael Crowley, PhD (Nov. 15, 2018).

have been revolutionary. The haploid human genome contains slightly more than three billion bases of DNA. When we inherit a single genome copy from our mother and one from our father, those two copies work together to form the diploid human genome. DNA sequence and organization are closely shared among humans with only small individual differences. The human genome project was a multinational effort completed in 2003 to generate a single reference genome that would support continued discovery in human genetics. The human reference genome has been under continuous update and improvement since 2003. Everyone has approximately three to four million variants compared to the reference genome with the exact number dependent on the ancestry and sex of the person. Those variants and differences in how the genome is organized define the genetic blueprint for each person. Our genome and unique collection of variants define how we look, our features, and the diseases we are susceptible to. Our genome along with our environment and lifestyle choices define our health and how we age and die. Although genetics contribute significantly to disease risk and progression, environment and lifestyle are also critical factors.

Advancements in technology over the last ten years have revolutionized the ability of scientists and clinicians to analyze how genetics influence health and disease. The completion of the human genome project provided the foundation of how to interpret changes in DNA sequence and organization. Today, comprehensive genetic testing, including whole-genome sequencing is widely accessible. International efforts have sequenced over one million genomes and several million samples have been analyzed using focused testing methods such as exome sequencing or targeted panel sequencing. Exome sequencing analyzes the portion of the human genome that encodes proteins, which is only ~2% of the total genome. Targeted panel sequencing is an approach where a small collection of genes known to be involved in a disease are analyzed. Exome and targeted panels have the advantage of being lower cost and easier to interpret compared to genome sequencing.

The collective knowledge from analyzing millions of samples has resulted in comprehensive resources for interpreting genetic testing results. When a genetic test is performed, the test results are compared to the reference genome. Variants from the reference genome are identified and annotated based on the collective knowledge. A consistent method and language to annotate variants has been developed. Variants directly involved in disease are identified as “Pathogenic Variants”. Variants that are known to be neutral are annotated as “Benign”. Genetic

testing results also identify variants that have uncertain significance. These variants usually alter a protein sequence, but that alteration has not been proven to be associated with disease. These are called VUS for “variants of unknown/uncertain significance”. The annotation status of a particular variant can be changed when there is sufficient evidence to support the change. VUS will commonly be advanced to either “Pathogenic” or “Likely Pathogenic” if there are multiple examples of that variant being detected with a specific disease. A VUS can also be annotated as “Benign” if the variant is frequently found in individuals with no evidence of disease.

Genetic testing is a common part of cancer treatment, and many companies offer tests of varying complexity. The range of complexity of these tests is from one gene to 600 genes. The larger panels that examine hundreds of genes are called pan-cancer tests since they analyze a collection of genes that are known or associated with cancer or cancer treatment but are not focused on a specific type of cancer. Single-gene tests such as sequencing BRCA1 and 2 are typically used when the analysis is focused on a specific type of cancer. These tests are intended to identify variants that alter treatment or prognosis based on annotations from previous studies. One of the limitations of focused testing in cancer is how to interpret VUS. The focused test is, by nature, biased to genes involved in cancer. When tests are negative for pathogenic variants, the next tier of interpretation is for VUS detected in the patient. It is standard practice to report VUS in genetic testing results. VUS may be further considered after further analysis such as examining the conservation of the variant or how frequently the variant has been observed in the total population of results. If a variant is observed in the population at a rate higher than the disease incidence, it is extremely unlikely that it is associated with the disease.

I have reviewed the genetic testing results of six plaintiffs. Each of them underwent genetic testing as part of their cancer treatment. The tests and interpretation were performed at different times to determine if there were known mutations that would alter the treatment of their disease or provide information valuable for family counseling. Each patient was provided with reports or a summary of results. This review is based on the content of those reports. None of the six patients had mutations in BRCA1 or 2 nor did they have known, pathogenic mutations that would influence treatment or prognosis. The testing and results for each of the patients are summarized below.

B. Patient Testing Results

1) Linda Bondurant

Ms. Bondurant was diagnosed with clear cell carcinoma.⁸ Testing was performed at MD Anderson where germline genetic testing identified a likely pathogenic variant of *SDHA* and a VUS in *PTCH1*.⁹ Screening of *BRCA1* and *BRCA2* was negative. No other details on the testing results were provided. *SDHA* encodes a subunit of succinate dehydrogenase, a mitochondrial enzyme involved in the Krebs cycle. Mutations in *SDHA* can lead to paraganglioma-pheochromocytoma syndrome (PGL1), characterized by an increased risk of paragangliomas and pheochromocytomas. These conditions were not observed in Ms. Bondurant, who was diagnosed with clear cell ovarian carcinoma.¹⁰ *PTCH1* is a tumor suppressor gene that plays a role in the Hedgehog signaling pathway, which is involved in cell growth and development. Mutations in *PTCH1* can lead to Gorlin syndrome, characterized by an increased risk of basal cell carcinoma, a type of skin cancer (Yang 2022). In my opinion, neither *SDHA* nor *PTCH1* have been implicated in ovarian cancer.

2) Hillary Converse

Ms. Converse was diagnosed with clear cell ovarian cancer.¹¹ Testing was performed on a blood sample at Myriad Genetics and was negative for pathogenic variants in *BRCA1* and *BRCA2*,¹² including negative for three variants found in people of Ashkenazi Jewish descent.¹³ Ms. Converse is of Jewish descent on both sides of her family and with a significant family history of cancer on her maternal side.¹⁴ Additional genetic testing was performed at Yale and a detailed report was provided. The variant interpretation from the report is quoted below:

“ATM, heterozygous variant. L2307F (MG19, chromosome 11:108196898 C>T). This variant has a frequency of 0.5%. It has not been reported as either a normal variant or a disease-causing mutation. The amino acid at the position is highly conserved, and the SIFT and Polyphen programs predict a deleterious effect of this variant. The significance of this variant should be evaluated in the clinical context.

⁸ BondurantL-MDAMR-00872; BondurantL-TCCMR-01667.

⁹ BondurantL-MDAMR-01167, BondurantL-MDAMR-01168.

¹⁰ BondurantL-MDAMR-01262, BondurantL-MDAMR-00872, BondurantL-MDAMR-01168.

¹¹ CONVERSE_HILARY_YALE_NEWHAVENHEALTH_000296.

¹² CONVERSE_HILARY_YALENEWHAVENHOSPITAL_01217.

¹³ CONVERSE_HILARY_YALENEWHAVENHOSPITAL_01215.

¹⁴ CONVERSE_HILARY_YALENEWHAVENHOSPITAL_01212.

Testing additional family members. if available, may be useful in evaluating the segregation of this variant with disease in the family.”¹⁵

Although this variant changes a conserved amino acid, reviewing this variant in ClinVar finds the majority of annotations are benign or likely benign without a report of association with ovarian cancer.^{16, 17}

A second variant was reported:

“TGFBk2, heterozygous variant, A329T (HG19, chromosome 3:30713680 G>A). This variant has a frequency of less than 1/10,000. It has been reported as causing Loeys Dietz syndrome, but also was seen in an unaffected member of a Loeys Diet Family, suggesting that it does not cause Loeys Diet syndrome (Looys (2006) NEngl J Med 355, 788). A different amino acid change in this gene at position 315 was reported in a single family with Lynch syndrome. This result has not been replicated in additional families. The amino acid at position 329 is not well conserved, and SIFT and Polyphen predict no deleterious effect. This variant is unlikely to be related to the patient's phenotype.”¹⁸

In my opinion, there is no evidence that either of these variants is pathogenic or has any association with an increased risk of ovarian cancer. These same two variants were found in Ms. Converse’s mother who developed breast cancer at age 46. The report notes that the precise role of these variants in cancer is not fully established but recommends Ms. Converse continue screening for breast cancer.¹⁹

3) Anna Gallardo

Ms. Gallardo was diagnosed with endometrioid ovarian cancer.²⁰ Testing was done on 11 genes through GeneDx Laboratory and no mutations were found.²¹ Due to these results, no additional testing was recommended for Ms. Gallardo or her relatives, as the screening she received “rules out the genes most commonly associated with hereditary risk for ovarian cancer.”²²

¹⁵ CONVERSE_HILARY_PROHEALTHPHYSICIANSOFHAMDEN_00297.

¹⁶ <https://www.ncbi.nlm.nih.gov/clinvar/variation/127430/>

¹⁷ CONVERSEH-FINEE-00075.

¹⁸ CONVERSE_HILARY_PROHEALTHPHYSICIANSOFHAMDEN_00297.

¹⁹ CONVERSE_HILARY_YALENEWHAVENHOSPITAL_01213.

²⁰ GALLARDO_A_BARNES_JEWISH_HOSP_000273; GALLARDO_ANNA_WASHU_MED-00324.

²¹ GALLARDO_A_GENEDX_000034 - GALLARDO_A_GENEDX_000036; GALLARDO_A_GENEDX_000123 - GALLARDO_A_GENEDX_000124.

²² GALLARDO_A_WASHU_000170.

4) Carter Judkins

Ms. Judkins was diagnosed with high grade serous ovarian cancer.²³ Testing was performed on a blood sample at Ambry Genetics, wherein a variant of unknown significance was found in *PTEN* (*c.-1283G>A*).²⁴ The OvaNext test analyzed 25 genes associated with hereditary ovarian cancer. The report stated “No known clinically actionable alterations were detected.”²⁵ The patient also underwent tumor testing at Myriad Genetics. The Myriad results were negative for *BRCA1* and *BRCA2* and positive for genomic instability in the tumor. Genomic instability is a prognostic marker of the tumor and may indicate a rapidly growing tumor or one that has multiple genetic changes. Genomic instability is observed in many cancers. Mutations in *PTEN* are associated with an increased risk of breast, endometrial, and thyroid cancer. *PTEN* is a tumor suppressor gene that plays a role in cell growth and apoptosis. There are over 1,000 variants in *PTEN* that have been annotated as pathogenic or likely pathogenic and 1,180 VUS. While the genetic testing report indicates that *PTEN* mutations in general are associated with susceptibility to ovarian cancer,²⁶ the specific mutation identified in Ms. Judkins has not been previously reported and in my opinion, there is no evidence to define it as a pathogenic variant or that there is any association with increased risk of ovarian cancer.

5) Tamara Newsome

Ms. Newsome was diagnosed with endometrioid adenocarcinoma of the ovary.²⁷ Blood sample testing was performed at Myriad Genetics using the MyRisk test and provided results on 25 genes. A single variant was reported as a VUS. *MUTYH c.1513T>G* (p.Cys505Gly).²⁸ *MUTYH* is a gene that encodes a protein involved in DNA repair. Mutations in *MUTYH* have been associated with a small increased risk of colorectal cancer, and monoallelic carriers reportedly have an increased risk for colorectal, liver, gastric and endometrial cancers, with conflicting findings with respect to breast cancer (Rodriguez-Rojas, Candelo et al. 2023). The region of *MUTYH* where the mutation is located is poorly conserved and adjacent sites are reported to be

²³ JUDKINSC_DHMC_C_MDR000085.

²⁴ JUDKINSC_REC000003.

²⁵ JUDKINSC_REC000003.

²⁶ JUDKINSC_REC000004.

²⁷ NEWSOMET_HCH_MDR000036.

²⁸ NEWSOMET_REC000003.

frequently mutated.²⁹ In my opinion, there is no evidence that this variant is pathogenic, nor that there is any association with an increased risk of ovarian cancer.

6) Pasqualina Rausa

Ms. Rausa was diagnosed with high grade serous carcinoma.³⁰ Testing was performed on a blood sample from Ms. Rausa with the Comprehensive Common Cancer test from GenPath. The test provides results on 46 genes with known involvement in cancer generally; it is not targeted toward ovarian cancer. One mutation was reported as a VUS in fumarate hydratase (*FH* c.986A>G).³¹ Mutations in *FH* can lead to hereditary leiomyomatosis and renal cell cancer (HLRCC), characterized by an increased risk of renal cell carcinoma. The genetic testing report points out that the specific mutation was found in at least one case of head and neck paraganglioma, but does not associate it with ovarian cancer.³² The report further points out the “variant is not located in a known functional domain,” indicating the mutation does not affect a site responsible for a function or interaction.³³ In my opinion, there is no additional evidence that this VUS is pathogenic or has an association with increased risk of ovarian cancer.

VIII. Conclusion

Based on my background, training, education, and experience as a geneticist assessing and weighing the totality of scientific and medical evidence, my opinions, which I hold to a reasonable degree of scientific certainty, may be summarized as follows:

1. Genetic mutations can be inherited or acquired. Both types are associated with cancer, including ovarian cancer.
2. Inflammation has been shown to play a vital role in epithelial ovarian cancer.
3. Talcum powder products cause chronic inflammation.
4. Talcum powder product-induced inflammation causes damage to the DNA, genetic mutation, genomic instability, and cell transformation.

²⁹ ClinVar report: <https://www.ncbi.nlm.nih.gov/clinvar/variation/238339/>

³⁰ PRAUSAPL-000001.

³¹ RausaP-GenPathMR-00005.

³² RausaP-GenPathMR-00006.

³³ RausaP-GenPathMR-00006.

5. The properties and constituents of talcum powder products act as inflammatory agents and the role of inflammation in triggering oxidative stress, activating cytokines, cell proliferation, DNA damage, and genetic mutations (such as SNVs) provide a biologically plausible mechanism for the carcinogenicity of talcum powder products.
6. Internalization of asbestiform fibers (including asbestos and fibrous talc) causes DNA damage, which provides a biologically plausible mechanism for the carcinogenicity of talcum powder products.
7. The presence of an inherited gene mutation, such as *BRCA1* or *BRCA2*, indicates a woman has an increased risk of ovarian cancer, but does not necessarily mean she will develop ovarian cancer.
8. Women with inherited gene mutations in genes involved in DNA repair, such as *BRCA1* or *BRCA2*, are more as susceptible to the effect of carcinogens than women without inherited gene mutations.
9. Genetic testing is a powerful and valuable tool to guide cancer treatment, prognosis, and family planning. All patients tested were negative for highly penetrant and clinically actionable mutations in *BRCA1* and *BRCA2*.
10. In relation to the VUS noted in the six patients reviewed, there is no evidence that the variants are pathogenic with regard to ovarian cancer or have any association with an increased risk of ovarian cancer.

I reserve the right to supplement, revise, or amend this report should additional materials, including expert reports and testimony, become available. I reserve the right to review and comment on defendants' expert reports and any related testimony.

Literature Cited

Adami, H. O., C. C. Hsieh, M. Lambe, D. Trichopoulos, D. Leon, I. Persson, A. Ekblom and P. O. Janson (1994). "Parity, age at first childbirth, and risk of ovarian cancer." Lancet **344**(8932): 1250-1254.

Agic, A., H. Xu, D. Finas, C. Banz, K. Diedrich and D. Hornung (2006). "Is endometriosis associated with systemic subclinical inflammation?" Gynecol Obstet Invest **62**(3): 139-147.

Andreassen, P. R., J. Seo, C. Wiek and H. Hanenberg (2021). "Understanding BRCA2 Function as a Tumor Suppressor Based on Domain-Specific Activities in DNA Damage Responses." Genes (Basel) **12**(7).

Banerjee, S. and S. B. Kaye (2013). "New strategies in the treatment of ovarian cancer: current clinical perspectives and future potential." Clin Cancer Res **19**(5): 961-968.

Barnes, D. R. and A. C. Antoniou (2012). "Unravelling modifiers of breast and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: update on genetic modifiers." J Intern Med **271**(4): 331-343.

Bayraktar, S., M. Jackson, A. M. Gutierrez-Barrera, D. Liu, F. Meric-Bernstam, A. Brandt, A. Woodson, J. Litton, K. H. Lu, V. Valero and B. K. Arun (2015). "Genotype-Phenotype Correlations by Ethnicity and Mutation Location in BRCA Mutation Carriers." Breast J **21**(3): 260-267.

Berek, J. S., C. Crum and M. Friedlander (2012). "Cancer of the ovary, fallopian tube, and peritoneum." Int J Gynaecol Obstet **119 Suppl 2**: S118-129.

Bodmer, M., C. Becker, C. Meier, S. S. Jick and C. R. Meier (2011). "Use of metformin and the risk of ovarian cancer: a case-control analysis." Gynecol Oncol **123**(2): 200-204.

Bonovas, S., K. Filioussi and N. M. Sitaras (2005). "Do nonsteroidal anti-inflammatory drugs affect the risk of developing ovarian cancer? A meta-analysis." Br J Clin Pharmacol **60**(2): 194-203.

Bowtell, D. D. (2010). "The genesis and evolution of high-grade serous ovarian cancer." Nat Rev Cancer **10**(11): 803-808.

Brieger, K. K., M. T. Phung, B. Mukherjee, K. M. Bakulski, H. Anton-Culver, E. V. Bandera, D. D. L. Bowtell, D. W. Cramer, A. DeFazio, J. A. Doherty, S. Fereday, R. T. Fortner, A. Gentry-Maharaj, E. L. Goode, M. T. Goodman, H. R. Harris, K. Matsuo, U. Menon, F. Modugno, K. B. Moysich, B. Qin, S. J. Ramus, H. A. Risch, M. A. Rossing, J. M. Schildkraut, B. Trabert, R. A. Vierkant, S. J. Winham, N. Wentzensen, A. H. Wu, A. Ziogas, L. Khoja, K. R. Cho, K. McLean, J. Richardson, B. Grout, A. Chase, C. M. Deurloo, K. Odunsi, B. H. Nelson, J. D. Brenton, K. L. Terry, P. D. P. Pharoah, A. Berchuck, G. E. Hanley, P. M. Webb, M. C. Pike, C. L. Pearce and C. Ovarian Cancer Association (2022). "High Prediagnosis Inflammation-Related Risk Score Associated with Decreased Ovarian Cancer Survival." Cancer Epidemiol Biomarkers Prev **31**(2): 443-452.

Buz'Zard, A. R. and B. H. Lau (2007). "Pycnogenol reduces talc-induced neoplastic transformation in human ovarian cell cultures." Phytother Res **21**(6): 579-586.

Camargo, M. C., L. T. Stayner, K. Straif, M. Reina, U. Al-Alem, P. A. Demers and P. J. Landrigan (2011). "Occupational exposure to asbestos and ovarian cancer: a meta-analysis." Environ Health Perspect **119**(9): 1211-1217.

Casagrande, J. T., E. W. Louie, M. C. Pike, S. Roy, R. K. Ross and B. E. Henderson (1979). "'Incessant ovulation' and ovarian cancer." Lancet **2**(8135): 170-173.

Chan, J. K., M. K. Cheung, A. Husain, N. N. Teng, D. West, A. S. Whittemore, J. S. Berek and K. Osann (2006). "Patterns and progress in ovarian cancer over 14 years." Obstet Gynecol **108**(3 Pt 1): 521-528.

Charbonneau, B., E. L. Goode, K. R. Kalli, K. L. Knutson and M. S. Derycke (2013). "The immune system in the pathogenesis of ovarian cancer." Crit Rev Immunol **33**(2): 137-164.

Coppa, A., A. Buffone, C. Capalbo, A. Nicolussi, S. D'Inzeo, F. Belardinilli, V. Colicchia, M. Petroni, T. Granato, C. Midulla, M. Zani, S. Ferraro, I. Screpanti, A. Gulino and G. Giannini (2014). "Novel and recurrent BRCA2 mutations in Italian breast/ovarian cancer families widen the ovarian cancer cluster region boundaries to exons 13 and 14." Breast Cancer Res Treat **148**(3): 629-635.

Coussens, L. M., C. L. Tinkle, D. Hanahan and Z. Werb (2000). "MMP-9 supplied by bone marrow-derived cells contributes to skin carcinogenesis." Cell **103**(3): 481-490.

Coussens, L. M. and Z. Werb (2002). "Inflammation and cancer." Nature **420**(6917): 860-867.

Emi, T., L. M. Rivera, V. C. Tripathi, N. Yano, A. Ragavendran, J. Wallace and A. V. Fedulov (2021). "Transcriptomic and epigenomic effects of insoluble particles on J774 macrophages." Epigenetics **16**(10): 1053-1070.

Fletcher, N. M., A. K. Harper, I. Memaj, R. Fan, R. T. Morris and G. M. Saed (2019). "Molecular Basis Supporting the Association of Talcum Powder Use With Increased Risk of Ovarian Cancer." Reprod Sci **26**(12): 1603-1612.

Forbes, S. A., D. Beare, P. Gunasekaran, K. Leung, N. Bindal, H. Boutselakis, M. Ding, S. Bamford, C. Cole, S. Ward, C. Y. Kok, M. Jia, T. De, J. W. Teague, M. R. Stratton, U. McDermott and P. J. Campbell (2015). "COSMIC: exploring the world's knowledge of somatic mutations in human cancer." Nucleic Acids Res **43**(Database issue): D805-811.

Franklin, R. A. and M. O. Li (2016). "Ontogeny of Tumor-associated Macrophages and Its Implication in Cancer Regulation." Trends Cancer **2**(1): 20-34.

Gabriel, I. M., A. F. Vitonis, W. R. Welch, L. Titus and D. W. Cramer (2019). "Douching, Talc Use, and Risk for Ovarian Cancer and Conditions Related to Genital Tract Inflammation." Cancer Epidemiol Biomarkers Prev **28**(11): 1835-1844.

Gates, M. A., S. S. Tworoger, K. L. Terry, L. Titus-Ernstoff, B. Rosner, I. De Vivo, D. W. Cramer and S. E. Hankinson (2008). "Talc use, variants of the GSTM1, GSTT1, and NAT2 genes, and risk of epithelial ovarian cancer." Cancer Epidemiol Biomarkers Prev **17**(9): 2436-2444.

Hamilton, T. C., H. Fox, C. H. Buckley, W. J. Henderson and K. Griffiths (1984). "Effects of talc on the rat ovary." Br J Exp Pathol **65**(1): 101-106.

Heller, D. S., R. E. Gordon, C. Westhoff and S. Gerber (1996). "Asbestos exposure and ovarian fiber burden." Am J Ind Med **29**(5): 435-439.

Hesterberg, T. W., C. J. Butterick, M. Oshimura, A. R. Brody and J. C. Barrett (1986). "Role of phagocytosis in Syrian hamster cell transformation and cytogenetic effects induced by asbestos and short and long glass fibers." Cancer Res **46**(11): 5795-5802.

Hurwitz, L. M., P. M. Webb, S. J. Jordan, J. A. Doherty, H. R. Harris, M. T. Goodman, Y. B. Shvetsov, F. Modugno, K. B. Moysich, J. M. Schildkraut, A. Berchuck, H. Anton-Culver, A. Ziogas, U. Menon, S. J. Ramus, A. H. Wu, C. L. Pearce, N. Wentzensen, S. S. Tworoger, P. D. P. Pharoah and B. Trabert (2023). "Association of Frequent Aspirin Use With Ovarian Cancer Risk According to Genetic Susceptibility." JAMA Netw Open **6**(2): e230666.

IARC (1987). Overall evaluations of carcinogenicity: an updating of IARC Monographs volumes 1 to 42.

IARC (1987). Silica and Some Silicates.

IARC (2006). Cobalt in Hard Metals and Cobalt Sulfate, Gallium Arsenide, Indium Phosphide and Vanadium Pentoxide.

IARC (2012). Arsenic, metals, fibres, and dusts.

Irie, H., K. Banno, M. Yanokura, M. Iida, M. Adachi, K. Nakamura, K. Umene, Y. Nogami, K. Masuda, Y. Kobayashi, E. Tominaga and D. Aoki (2016). "Metformin: A candidate for the treatment of gynecological tumors based on drug repositioning." Oncol Lett **11**(2): 1287-1293.

Jayson, G. C., E. C. Kohn, H. C. Kitchener and J. A. Ledermann (2014). "Ovarian cancer." Lancet **384**(9951): 1376-1388.

Jia, D., Y. Nagaoka, M. Katsumata and S. Orsulic (2018). "Inflammation is a key contributor to ovarian cancer cell seeding." Sci Rep **8**(1): 12394.

Jordan, S. J., K. L. Cushing-Haugen, K. G. Wicklund, J. A. Doherty and M. A. Rossing (2012). "Breast-feeding and risk of epithelial ovarian cancer." Cancer Causes Control **23**(6): 919-927.

Kadry Taher, M., N. Farhat, N. A. Karyakina, N. Shilnikova, S. Ramoju, C. A. Gravel, K. Krishnan, D. Mattison, S. W. Wen and D. Krewski (2019). "Critical review of the association between perineal use of talc powder and risk of ovarian cancer." Reprod Toxicol **90**: 88-101.

Keskin, N., Y. A. Teksen, E. G. Ongun, Y. Ozay and H. Saygili (2009). "Does long-term talc exposure have a carcinogenic effect on the female genital system of rats? An experimental pilot study." Arch Gynecol Obstet **280**(6): 925-931.

Kisielewski, R., A. Tolwinska, A. Mazurek and P. Laudanski (2013). "Inflammation and ovarian cancer--current views." Ginekol Pol **84**(4): 293-297.

Langseth, H., B. V. Johansen, J. M. Nesland and K. Kjaerheim (2007). "Asbestos fibers in ovarian tissue from Norwegian pulp and paper workers." Int J Gynecol Cancer **17**(1): 44-49.

Levanon, K., C. Crum and R. Drapkin (2008). "New insights into the pathogenesis of serous ovarian cancer and its clinical impact." J Clin Oncol **26**(32): 5284-5293.

Li, D. (2011). "Metformin as an antitumor agent in cancer prevention and treatment." J Diabetes **3**(4): 320-327.

Lin, H. W., Y. Y. Tu, S. Y. Lin, W. J. Su, W. L. Lin, W. Z. Lin, S. C. Wu and Y. L. Lai (2011). "Risk of ovarian cancer in women with pelvic inflammatory disease: a population-based study." Lancet Oncol **12**(9): 900-904.

Liu, Y. and X. Cao (2015). "The origin and function of tumor-associated macrophages." Cell Mol Immunol **12**(1): 1-4.

Longo, W. and M. Rigler (2017). Analysis of Johnson & Johnson Baby Powder & Valiant Shower to Shower Talc Products for Amphibole (Tremolite) Asbestos, Materials Analytical Services, LLC.

Longo, W. and M. Rigler (2018). TEM Analysis of Historical 1978 Johnson's Baby Powder Sample for Amphibole Asbestos, Materials Analytical Services, LLC.

Mabuchi, S., C. Kawase, D. A. Altomare, K. Morishige, K. Sawada, M. Hayashi, M. Tsujimoto, M. Yamoto, A. J. Klein-Szanto, R. J. Schilder, M. Ohmichi, J. R. Testa and T. Kimura (2009). "mTOR is a promising therapeutic target both in cisplatin-sensitive and cisplatin-resistant clear cell carcinoma of the ovary." Clin Cancer Res **15**(17): 5404-5413.

Maccio, A. and C. Madeddu (2012). "Inflammation and ovarian cancer." Cytokine **58**(2): 133-147.

Mahdavi, A., T. Pejovic and F. Nezhat (2006). "Induction of ovulation and ovarian cancer: a critical review of the literature." Fertil Steril **85**(4): 819-826.

Mandarino, A., D. J. Gregory, C. C. McGuire, B. W. Leblanc, H. Witt, L. M. Rivera, J. J. Godleski and A. V. Fedulov (2020). "The effect of talc particles on phagocytes in co-culture with ovarian cancer cells." Environ Res **180**: 108676.

Mantovani, A., B. Bottazzi, F. Colotta, S. Sozzani and L. Ruco (1992). "The origin and function of tumor-associated macrophages." Immunol Today **13**(7): 265-270.

Mantovani, A., F. Bussolino and E. Dejana (1992). "Cytokine regulation of endothelial cell function." FASEB J **6**(8): 2591-2599.

Mantovani, A., F. Bussolino and M. Introna (1997). "Cytokine regulation of endothelial cell function: from molecular level to the bedside." Immunol Today **18**(5): 231-240.

Merritt, M. A., A. C. Green, C. M. Nagle, P. M. Webb, S. Australian Cancer and G. Australian Ovarian Cancer Study (2008). "Talcum powder, chronic pelvic inflammation and NSAIDs in relation to risk of epithelial ovarian cancer." Int J Cancer **122**(1): 170-176.

Mills, P. K., D. G. Riordan, R. D. Cress and H. A. Young (2004). "Perineal talc exposure and epithelial ovarian cancer risk in the Central Valley of California." Int J Cancer **112**(3): 458-464.

Modan, B., P. Hartge, G. Hirsh-Yechezkel, A. Chetrit, F. Lubin, U. Beller, G. Ben-Baruch, A. Fishman, J. Menczer, J. P. Struewing, M. A. Tucker, S. Wacholder and G. National Israel Ovarian Cancer Study (2001). "Parity, oral contraceptives, and the risk of ovarian cancer among carriers and noncarriers of a BRCA1 or BRCA2 mutation." N Engl J Med **345**(4): 235-240.

Mor, G., G. Yin, I. Chetetz, Y. Yang and A. Alvero (2011). "Ovarian cancer stem cells and inflammation." Cancer Biol Ther **11**(8): 708-713.

Narod, S. A., H. Risch, R. Moslehi, A. Dorum, S. Neuhausen, H. Olsson, D. Provencher, P. Radice, G. Evans, S. Bishop, J. S. Brunet and B. A. Ponder (1998). "Oral contraceptives and the risk of hereditary ovarian cancer. Hereditary Ovarian Cancer Clinical Study Group." N Engl J Med **339**(7): 424-428.

National Toxicology, P. (1993). "NTP Toxicology and Carcinogenesis Studies of Talc (CAS No. 14807-96-6)(Non-Asbestiform) in F344/N Rats and B6C3F1 Mice (Inhalation Studies)." Natl Toxicol Program Tech Rep Ser **421**: 1-287.

Ness, R. B., J. A. Grisso, C. Cottreau, J. Klapper, R. Vergona, J. E. Wheeler, M. Morgan and J. J. Schlesselman (2000). "Factors related to inflammation of the ovarian epithelium and risk of ovarian cancer." Epidemiology **11**(2): 111-117.

Nunes, S. C. and J. Serpa (2018). "Glutathione in Ovarian Cancer: A Double-Edged Sword." Int J Mol Sci **19**(7).

O'Brien, K. M., A. A. D'Aloisio, M. Shi, J. D. Murphy, D. P. Sandler and C. R. Weinberg (2019). "Perineal Talc Use, Douching, and the Risk of Uterine Cancer." Epidemiology **30**(6): 845-852.

Paluch-Shimon, S., F. Cardoso, C. Sessa, J. Balmana, M. J. Cardoso, F. Gilbert, E. Senkus and E. G. Committee (2016). "Prevention and screening in BRCA mutation carriers and other breast/ovarian hereditary cancer syndromes: ESMO Clinical Practice Guidelines for cancer prevention and screening." Ann Oncol **27**(suppl 5): v103-v110.

Pardoll, D. M. (2002). "Spinning molecular immunology into successful immunotherapy." Nat Rev Immunol **2**(4): 227-238.

Park, H. K., J. M. Schildkraut, A. J. Alberg, E. V. Bandera, J. S. Barnholtz-Sloan, M. Bondy, S. Crankshaw, E. Funkhouser, P. G. Moorman, E. S. Peters, P. Terry, F. Wang, J. J. Ruterbusch, A. G. Schwartz and M. L. Cote (2018). "Benign gynecologic conditions are associated with ovarian cancer risk in African-American women: a case-control study." Cancer Causes Control **29**(11): 1081-1091.

Pejovic, T. and F. Nezhat (2011). "Missing link: inflammation and ovarian cancer." Lancet Oncol **12**(9): 833-834.

Penninkilampi, R. and G. D. Eslick (2018). "Perineal Talc Use and Ovarian Cancer: A Systematic Review and Meta-Analysis." Epidemiology **29**(1): 41-49.

Ramus, S. J., R. A. Vierkant, S. E. Johnatty, M. C. Pike, D. J. Van Den Berg, A. H. Wu, C. L. Pearce, U. Menon, A. Gentry-Maharaj, S. A. Gayther, R. A. DiCioccio, V. McGuire, A. S. Whittemore, H. Song, D. F. Easton, P. D. P. Pharoah, M. Garcia-Closas, S. Chanock, J. Lissowska, L. Brinton, K. L. Terry, D. W. Cramer, S. S. Tworoger, S. E. Hankinson, A. Berchuck, P. G. Moorman, J. M. Schildkraut, J. M. Cunningham, M. Liebow, S. K. Kjaer, E. Hogdall, C. Hogdall, J. Blaakaer, R. B. Ness, K. B. Moysich, R. P. Edwards, M. E. Carney, G. Lurie, M. T. Goodman, S. Wang-Gohrke, S. Kropp, J. Chang-Claude, P. M. Webb, X. Chen, J. Beesley, G. Chenevix-Trench and E. L. Goode (2008). "Consortium analysis of 7 candidate SNPs for ovarian cancer." Int J Cancer **123**(2): 380-388.

Reid, B. M., J. B. Permeth and T. A. Sellers (2017). "Epidemiology of ovarian cancer: a review." Cancer Biol Med **14**(1): 9-32.

Rodriguez-Rojas, L. X., E. Candelo, H. Pachajoa, J. E. Garcia-Robledo, J. A. Nastasi-Catanese, J. A. Olave-Rodriguez and A. R. Zambrano (2023). "The Unique Spectrum of MUTYH Germline Mutations in Colombian Patients with Extracolonic Carcinomas." Appl Clin Genet **16**: 53-62.

Rosenblatt, K. A., N. S. Weiss, K. L. Cushing-Haugen, K. G. Wicklund and M. A. Rossing (2011). "Genital powder exposure and the risk of epithelial ovarian cancer." Cancer Causes Control **22**(5): 737-742.

Saed, G. M., M. P. Diamond and N. M. Fletcher (2017). "Updates of the role of oxidative stress in the pathogenesis of ovarian cancer." Gynecol Oncol **145**(3): 595-602.

Sellers, T. A., Y. Huang, J. Cunningham, E. L. Goode, R. Sutphen, R. A. Vierkant, L. E. Kelemen, Z. S. Fredericksen, M. Liebow, V. S. Pankratz, L. C. Hartmann, J. Myer, E. S. Iversen, Jr., J. M. Schildkraut and C. Phelan (2008). "Association of single nucleotide polymorphisms in glycosylation genes with risk of epithelial ovarian cancer." Cancer Epidemiol Biomarkers Prev **17**(2): 397-404.

Shan, W. and J. Liu (2009). "Inflammation: a hidden path to breaking the spell of ovarian cancer." Cell Cycle **8**(19): 3107-3111.

Shen, H., B. L. Fridley, H. Song, K. Lawrenson, J. M. Cunningham, S. J. Ramus, M. S. Cicek, J. Tyrer, D. Stram, M. C. Larson, M. Kobel, P. Consortium, A. Ziogas, W. Zheng, H. P. Yang, A.

H. Wu, E. L. Wozniak, Y. L. Woo, B. Winterhoff, E. Wik, A. S. Whittemore, N. Wentzensen, R. P. Weber, A. F. Vitonis, D. Vincent, R. A. Vierkant, I. Vergote, D. Van Den Berg, A. M. Van Altena, S. S. Tworoger, P. J. Thompson, D. C. Tessier, K. L. Terry, S. H. Teo, C. Templeman, D. O. Stram, M. C. Southey, W. Sieh, N. Siddiqui, Y. B. Shvetsov, X. O. Shu, V. Shridhar, S. Wang-Gohrke, G. Severi, I. Schwaab, H. B. Salvesen, I. K. Rzepecka, I. B. Runnebaum, M. A. Rossing, L. Rodriguez-Rodriguez, H. A. Risch, S. P. Renner, E. M. Poole, M. C. Pike, C. M. Phelan, L. M. Pelttari, T. Pejovic, J. Paul, I. Orlow, S. Z. Omar, S. H. Olson, K. Odunsi, S. Nickels, H. Nevanlinna, R. B. Ness, S. A. Narod, T. Nakanishi, K. B. Moysich, A. N. Monteiro, J. Moes-Sosnowska, F. Modugno, U. Menon, J. R. McLaughlin, V. McGuire, K. Matsuo, N. A. Adenan, L. F. Massuger, G. Lurie, L. Lundvall, J. Lubinski, J. Lissowska, D. A. Levine, A. Leminen, A. W. Lee, N. D. Le, S. Lambrechts, D. Lambrechts, J. Kupryjanczyk, C. Krakstad, G. E. Konecny, S. K. Kjaer, L. A. Kiemeny, L. E. Kelemen, G. L. Keeney, B. Y. Karlan, R. Karevan, K. R. Kalli, H. Kajiyama, B. T. Ji, A. Jensen, A. Jakubowska, E. Iversen, S. Hosono, C. K. Hogdall, E. Hogdall, M. Hoatlin, P. Hillemanns, F. Heitz, R. Hein, P. Harter, M. K. Halle, P. Hall, J. Gronwald, M. Gore, M. T. Goodman, G. G. Giles, A. Gentry-Maharaj, M. Garcia-Closas, J. M. Flanagan, P. A. Fasching, A. B. Ekici, R. Edwards, D. Eccles, D. F. Easton, M. Durst, A. du Bois, T. Dork, J. A. Doherty, E. Despierre, A. Dansonka-Mieszkowska, C. Cybulski, D. W. Cramer, L. S. Cook, X. Chen, B. Charbonneau, J. Chang-Claude, I. Campbell, R. Butzow, C. H. Bunker, D. Brueggmann, R. Brown, A. Brooks-Wilson, L. A. Brinton, N. Bogdanova, M. S. Block, E. Benjamin, J. Beesley, M. W. Beckmann, E. V. Bandera, L. Baglietto, F. Bacot, S. M. Armasu, N. Antonenkova, H. Anton-Culver, K. K. Aben, D. Liang, X. Wu, K. Lu, M. A. Hildebrandt, G. Australian Ovarian Cancer Study, S. Australian Cancer, J. M. Schildkraut, T. A. Sellers, D. Huntsman, A. Berchuck, G. Chenevix-Trench, S. A. Gayther, P. D. Pharoah, P. W. Laird, E. L. Goode and C. L. Pearce (2013). "Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer." Nat Commun **4**: 1628.

Shukla, A., M. B. MacPherson, J. Hillegass, M. E. Ramos-Nino, V. Alexeeva, P. M. Vacek, J. P. Bond, H. I. Pass, C. Steele and B. T. Mossman (2009). "Alterations in gene expression in human mesothelial cells correlate with mineral pathogenicity." Am J Respir Cell Mol Biol **41**(1): 114-123.

Straif, K., L. Benbrahim-Tallaa, R. Baan, Y. Grosse, B. Secretan, F. El Ghissassi, V. Bouvard, N. Guha, C. Freeman, L. Galichet, V. Coglianò and W. H. O. I. A. f. R. o. C. M. W. Group (2009). "A review of human carcinogens--Part C: metals, arsenic, dusts, and fibres." Lancet Oncol **10**(5): 453-454.

Thompson, H. S., H. B. Valdimarsdottir, C. Duteau-Buck, J. Guevarra, D. H. Bovbjerg, C. Richmond-Avellaneda, D. Amarel, D. Godfrey, K. Brown and K. Offit (2002). "Psychosocial predictors of BRCA counseling and testing decisions among urban African-American women." Cancer Epidemiol Biomarkers Prev **11**(12): 1579-1585.

Titus-Ernstoff, L., K. Perez, D. W. Cramer, B. L. Harlow, J. A. Baron and E. R. Greenberg (2001). "Menstrual and reproductive factors in relation to ovarian cancer risk." Br J Cancer **84**(5): 714-721.

Toland, A. E., L. C. Brody and B. I. C. S. Committee (2019). "Lessons learned from two decades of BRCA1 and BRCA2 genetic testing: the evolution of data sharing and variant classification." Genet Med **21**(7): 1476-1480.

Trabert, B., R. B. Ness, W. H. Lo-Ciganic, M. A. Murphy, E. L. Goode, E. M. Poole, L. A. Brinton, P. M. Webb, C. M. Nagle, S. J. Jordan, A. C. S. Australian Ovarian Cancer Study Group, H. A. Risch, M. A. Rossing, J. A. Doherty, M. T. Goodman, G. Lurie, S. K. Kjaer, E. Hogdall, A. Jensen, D. W. Cramer, K. L. Terry, A. Vitonis, E. V. Bandera, S. Olson, M. G. King, U. Chandran, H. Anton-Culver, A. Ziogas, U. Menon, S. A. Gayther, S. J. Ramus, A. Gentry-Maharaj, A. H. Wu, C. L. Pearce, M. C. Pike, A. Berchuck, J. M. Schildkraut, N. Wentzensen and C. Ovarian Cancer Association (2014). "Aspirin, nonaspirin nonsteroidal anti-inflammatory drug, and acetaminophen use and risk of invasive epithelial ovarian cancer: a pooled analysis in the Ovarian Cancer Association Consortium." J Natl Cancer Inst **106**(2): djt431.

Vitonis, A. F., L. Titus-Ernstoff and D. W. Cramer (2011). "Assessing ovarian cancer risk when considering elective oophorectomy at the time of hysterectomy." Obstet Gynecol **117**(5): 1042-1050.

Wang, N. S., M. C. Jaurand, L. Magne, L. Kheuang, M. C. Pinchon and J. Bignon (1987). "The interactions between asbestos fibers and metaphase chromosomes of rat pleural mesothelial cells in culture. A scanning and transmission electron microscopic study." Am J Pathol **126**(2): 343-349.

Wehner, A. P. (1994). "Biological effects of cosmetic talc." Food Chem Toxicol **32**(12): 1173-1184.

Wentzensen, N. and K. M. O'Brien (2021). "Talc, body powder, and ovarian cancer: A summary of the epidemiologic evidence." Gynecol Oncol **163**(1): 199-208.

Wishart, D. S. (2015). "Is Cancer a Genetic Disease or a Metabolic Disease?" EBioMedicine **2**(6): 478-479.

Woolen, S. A., A. A. Lazar and R. Smith-Bindman (2022). "Association Between the Frequent Use of Perineal Talcum Powder Products and Ovarian Cancer: a Systematic Review and Meta-analysis." J Gen Intern Med **37**(10): 2526-2532.

Wu, A. H., C. L. Pearce, C. C. Tseng and M. C. Pike (2015). "African Americans and Hispanics Remain at Lower Risk of Ovarian Cancer Than Non-Hispanic Whites after Considering Nongenetic Risk Factors and Oophorectomy Rates." Cancer Epidemiol Biomarkers Prev **24**(7): 1094-1100.

Wu, A. H., C. L. Pearce, C. C. Tseng, C. Templeman and M. C. Pike (2009). "Markers of inflammation and risk of ovarian cancer in Los Angeles County." Int J Cancer **124**(6): 1409-1415.

Yegles, M., L. Saint-Etienne, A. Renier, X. Janson and M. C. Jaurand (1993). "Induction of metaphase and anaphase/telophase abnormalities by asbestos fibers in rat pleural mesothelial cells in vitro." Am J Respir Cell Mol Biol **9**(2): 186-191.

Exhibit A

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Personal Statement

My research group has utilized high-performance genotyping and sequencing technologies for the past 20 years, supporting various projects from plant and animal phylogenetic studies to translational and clinical-based projects. Several publications detail our successes using various genomic technologies and in bioinformatics research. As a post-doctoral fellow at Emory University, I developed the first microarray to interrogate mitochondrial gene function. Upon joining the faculty at Vanderbilt University, I was responsible for founding and developing the Vanderbilt Microarray Shared Resource (VMSR). From 2000 to 2009, the VMSR became an internationally recognized facility supporting various genomic technologies from SNP profiling to gene expression analysis to next-generation sequencing. I joined the faculty of the HudsonAlpha Institute for Biotechnology in 2009 to develop the Genomic Services Laboratory (GSL). From 2009 through 2019, the GSL supported more than 6,000 projects from principal investigators worldwide. These projects have allowed me to collaborate and participate in various genomics projects focusing on genomic methods to decipher complex conditions. We have had a particular focus on childhood and adult cancer as well as rare disease and degenerative diseases. Our efforts have been both financially successful as well as academically successful. Our team has contributed to more than 200 peer-reviewed publications of which I am an author or co-author. More than 250 additional publications that have included data from our laboratory as a service provider have also been published since 2009. Many of these publications involve translational research or describe the genetic underpinnings of rare or complex human diseases. Although located in a non-profit institute, the GSL laboratory was developed with a focus on quality, efficiency, and innovation. Our financial and scientific successes and approach led to HudsonAlpha selling the assets of the GSL to Discovery Life Sciences in August 2019, creating a new division in the company called HudsonAlpha Discovery. The sale was the largest financial transaction in the institute's history and continues the legacy of the GSL through clinical and basic research support for academic and biopharmaceutical partners. The diversity of projects, technologies, and investigators we have worked and collaborated with over the last two decades have provided a dynamic and unique experience to evolve our own research and technology development efforts to help solve the most pressing challenges in biology. In 2022 I transitioned from Chief Scientific Officer of Discovery Life Sciences to a position at Element Biosciences. I currently serve as the Chief Scientific Officer of Element Biosciences and SVP-Applications. My responsibilities at Element Biosciences include the overall scientific strategy of the company as well as overseeing all applications development and collaborations.

Contributions to Science

The following five sections provide highlights to areas where my work has contributed to areas of science. Example publications are provided with each section and a full bibliography is provided at the end of the CV.

1. My scientific career has been a somewhat atypical in that I have spent the last 15 years focusing on the development and application of genomic and bioinformatic technologies and methods to support scientific investigation in a number of areas. While there have been substantial areas of focus, my laboratory does not operate under a single or specific biological area or hypothesis. Instead, we examine ways to improve the resolution and quality of results to answer complex questions, regardless of biological relationship. The publications below are examples of contributions to technical projects or large consortium projects with goals in the evaluation or improvement of techniques or technologies.
 - a. Statnikov A, Aliferis, C, Tsamardinos, I, Hardin, D, and Levy, S. A comprehensive evaluation of multicategory classification methods for microarray gene expression cancer diagnosis. **Bioinformatics**, 2005. 21(5), p. 631-643. PMID:15374862.
 - b. The MicroArray Quality Control Consortium. The MicroArray Quality Control (MAQC) project shows inter- and intraplatform reproducibility of gene expression measurements. **Nature Biotechnology**, 2006. 24(9), p. 1151-1161. PMID:16964229.
 - c. The ENCODE Project Consortium. An integrated encyclopedia of DNA elements in the human genome. **Nature**. 2012. 489, 57-74. PMID: 22955616 PMCID: PMC3439153
 - d. The Sequence Quality Control (SEQC) Consortium. A comprehensive assessment of RNA-seq accuracy, reproducibility and information content by the Sequence Quality Control consortium. **Nature Biotechnology**. 2014. 32 (9), 915-925. PMID:25150835; PMCID:4167418.

2. One area of early focus of my career was the development and analysis of mouse models for mitochondrial disease, including the knock out of the Adenine Nucleotide Translocase 2 (Ant2) gene leading to a more complete understanding of the permeability transition. This work also discovered methods to alter the mitochondrial DNA in stem cells and supported the first mitochondrial DNA transfers by stem cells.
 - a. Levy SE, Waymire, KG, Kim, YL, MacGregor, GR, and Wallace, DC, Transfer of chloramphenicol-resistant mitochondrial DNA into the chimeric mouse. **Transgenic Research**. 1999. 8(2), p. 137-145. PMID:10481313.
 - b. Sligh JE, Levy SE, Waymire KG, Allard P, Dillehay DL, Nusinowitz S, Heckenlively JR, MacGregor GR, and Wallace DC. Maternal germ-line transmission of mutant mtDNAs from embryonic stem cell-derived chimeric mice. **Proc. of the Nat. Acad. of Sciences USA**. 2000. 97(26), p. 14461-14466. PMID:11106380; PMCID:18941.
 - c. Kokoszka JE, Waymire, KG, Levy, SE, Sligh, JE, Cal, JY, Jones, DP, MacGregor, GR, and Wallace, DC, The ADP/ATP translocator is not essential for the mitochondrial permeability transition pore. **Nature**, 2004. 427(6973),p. 461-465. PMID:14749836.
 - d. Picard M, Zhang J, Hanecock S, Derbeneva O, Golhar R, Golik P, O'Hearn S, Levy SE, Potluri P, Lvova M, Davila A, Lin CS, Perin JC, Rappaport EF, Hakonarson H, Trounce I, Procaccio V, and Wallace DC. Progressive increase in mtDNA 3243A>G heteroplasmy results in abrupt transcriptional remodeling. **Proc. of the Nat. Acad. of Sciences USA**. 2014. 111(38), E4033-E4042. PMID:25192935; PMCID:4183335.

3. A long-standing area of research interest is the genomic analysis of cancer, both childhood

and adult. These efforts have included population-based studies and more directed research in specific cancer biology. These efforts have examined many cancer types including breast, lung, colon, and myeloid cancer.

- a. Smith JJ, Deane, NG, Wu, F, Merchant, NB, Zhang, B, Jiang, A, Lu, P, Johnson, JC, Schmidt, C, Edwards, CM, Eschrich, S, Kis, C, Levy, S, Washington, MK, Heslin, MJ, Coffey, RJ, Yeatman, TJ, Shyr, Y, and Beauchamp, RD, Experimentally Derived Metastasis Gene Expression Profile Predicts Recurrence and Death in Patients With Colon Cancer. **Gastroenterology**, 2009. PMID: 19914252 PMCID: PMC3388775.
 - b. Powell AE, Wang Y, Li Y, Poulin EJ, Means AL, Washington MK, Higginbotham JN, Juchheim A, Prasad N, Levy SE, Guo Y, Shyr Y, Aronow BJ, Haigis KM, Franklin JL, and Coffey RJ. Lrig1, a pan-ErbB negative regulator, marks intestinal stem cells and acts as a tumor suppressor. **Cell**. 2012. 149(1), 146-158. PMID: 22464327 PMCID: PMC3563328.
 - c. McDaniel JM, Varley KE, Gertz J, Savic DS, Roberts BS, Bailey SK, Shevde LA, Ramaker RC, Lasseigne BN, Kirby MK, Newberry KM, Partridge EC, Jones AL, Boone B, Levy SE, Oliver PG, Sexton KC, Grizzle WE, Forero A, Buchsbaum DJ, Cooper SJ, Myers RM. Genomic regulation of invasion by STAT3 in triple negative breast cancer. **Oncotarget**. 2017;8(5):8226-38. doi: 10.18632/oncotarget.14153. PubMed PMID: 28030809; PMCID: PMC5352396.
 - d. McKinney M, Moffitt AB, Gaulard P, Travert M, De Leval L, Nicolae A, Raffeld M, Jaffe ES, Pittaluga S, Xi L, Heavican T, Iqbal J, Belhadj K, Delfau-Larue MH, Fataccioli V, Czader MB, Lossos IS, Chapman-Fredricks JR, Richards KL, Fedoriw Y, Ondrejka SL, Hsi ED, Low L, Weisenburger D, Chan WC, Mehta-Shah N, Horwitz S, Bernal-Mizrachi L, Flowers CR, Beaven AW, Parihar M, Baseggio L, Parrens M, Moreau A, Sujobert P, Pilichowska M, Evens AM, Chadburn A, Au-Yeung RK, Srivastava G, Choi WW, Goodlad JR, Aurer I, Basic-Kinda S, Gascoyne RD, Davis NS, Li G, Zhang J, Rajagopalan D, Reddy A, Love C, Levy S, Zhuang Y, Datta J, Dunson DB, Dave SS. The Genetic Basis of Hepatosplenic T-cell Lymphoma. **Cancer Discov**. 2017;7(4):369-79. doi: 10.1158/2159-8290.CD-16-0330. PubMed PMID: 28122867; PMCID: PMC5402251.
4. My laboratory has had the opportunity to collaborate with a number of outstanding investigators in the genetics analysis of complex neurological conditions, including autism, schizophrenia and bipolar disorders as well as ALS. We contributed significantly to the discovery of the association of de-novo rather than Mendelian mutations in these conditions, particularly in schizophrenia.
- a. Xu B, Roos JL, Dexheimer P, Boone B, Plummer B, Levy S, Gogos JA, Karayiorgou M. Exome sequencing supports a de novo mutational paradigm for schizophrenia. **Nature Genetics**. 2011. 43(9), 864-868. PMID: 21822266. PMCID: PMC3196550.
 - b. Neale BM, Kou Y, Liu L, Ma'ayan A, Samocha KE, Sabo A, Lin CF, Stevens C, Wang LS, Makarov V, Polak P, Yoon S, Maguire J, Crawford EL, Campbell NG, Geller ET, Valladares O, Shafer C, Liu H, Zhao T, Cai G, Lihm J, Dannenfelser R, Jabado O, Peralta Z, Nagaswamy U, Reid JG, Newsham I, Wu Y, Lewis L, Han Y, Muzny D, Voight BF, Lim E, Rossin E, Kirby A, Flannick J, Fromer M, Shakir K, Fennell T, Garimella K, Boyko C, Gabriel S, dePristo M, Wimbish JR, Boone BE, Levy SE, Betancur C, Sunyaev S, Boerwinkle E, Buxbaum JD, Cook EH, Devlin B, Gibbs R, Roeder K, Schellenberg GD, Sutcliffe JS, and Daly MJ. Patterns and rates of exonic de novo mutations in autism spectrum disorders. **Nature**. 2012. 485(7397), 242-245. PMID: 22495311 PMCID:PMC3613847.
 - c. Xu B, Ionita-Laza I, Roos JL, Boone B, Woodrick S, Sun Y, Levy S, Gogos JA, and Karayiorgou M. De novo gene mutations highlight patterns of genetic and neural complexity

- in schizophrenia. **Nature Genetics**. 2012. 44(12), 1365-1369. PMID: 23042115 PMCID: PMC3556813.
- d. Cirulli, ET, Lasseigne, BN, Petrovski, S, Sapp, PC, Dion, PA, Leblond, CS, Couthouis, J, Lu, Y-F, Wang, Q, Krueger, BJ, Ren, Z, Keebler, J, Han, Y, Levy, SE, Boone, BE, Wimbish, JR, Waite, LL, Jones, AL, Carulli, JP, Day-Williams, AG, Staropoli, JF, Xin, WW, Chesi, A, Raphael, AR, McKenna-Yasek, D, Cady, J, Vianney de Jong, JMB, Kenna, KP, Smith, BN, Topp, S, Miller, J, Gkazi, A, Consortium, FS, Al-Chalabi, A, van den Berg, LH, Veldink, J, Silani, V, Ticozzi, N, Shaw, CE, Baloh, RH, Appel, S, Simpson, E, Lagier-Tourenne, C, Pulst, SM, Gibson, S, Trojanowski, JQ, Elman, L, McCluskey, L, Grossman, M, Shneider, NA, Chung, WK, Ravits, JM, Glass, JD, Sims, KB, Van Deerlin, VM, Maniatis, T, Hayes, SD, Ordureau, A, Swarup, S, Landers, J, Baas, F, Allen, AS, Bedlack, RS, Harper, JW, Gitler, AD, Rouleau, GA, Brown, R, Harms, MB, Cooper, GM, Harris, T, Myers, RM, Goldstein, DB. Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. **Science**. 2015. 347(6229): p. 1436-41.
 5. My laboratory has played a significant role in the discovery of the causative mutations of a number of rare but significant human diseases, particularly in the field of pediatric nephrology in collaboration with Friedhelm Hildebrandt at Harvard University. These studies applied genomic technologies to better characterize and, in some cases, diagnose or discover the causative mutation for severe phenotypes or disease.
 - a. Otto EA, Hurd TW, Airik R, Chaki M, Zhou W, Stoetzel C, Patil SB, Levy S, Ghosh AK, Murga-Zamalloa CA, van Reeuwijk J, Letteboer SJF, Sang L, Giles RH, Liu Q, Coene KLM, Estrada-Cuzcano A, Collin RWJ, McLaughlin HM, Held S, Kasanuki JM, Ramaswami G, Conte J, Lopez I, Washburn J, MacDonald J, Hu, J, Yamashita Y, Maher ER, Guay-Woodford L, Neumann HPH, Obermuller H, Koenekoop RK, Bergmann C, Bei X, Lewis RA, Katsanis N, Lopes V, Williams DS, Lyons RH, Dang CV, Brito DA, Dias MB, Zhang X, Nurnberg G, Nurnberg P, Pierce E, Jackson P, Antignac C, Saunier S, Roepman R, Dollfus H, Khanna H, and Hildebrandt F. Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. **Nature Genetics**, 2010. 42(10), 840-850 PMID: 20835237 PMCID: PMC2947620.
 - b. Rademakers R, Baker M, Nicholson AM, Rutherford NJ, Finch N, Soto-Ortolaza A, Lash J, Wider C, Wojtas A, DeJesus-Hernandez M, Adamson J, Kouri N, Sundal C, Shuster EA, Aasly J, MacKenzie J, Roeber S, Kretzschmar HA, Boeve BF, Knopman DS, Petersen RC, Cairns NJ, Ghetti B, Spina S, Garbern J, Tselis AC, Uitti R, Das P, Van Gerpen JA, Meschia JF, Levy S, Broderick DF, Graff-Radford N, Ross OA, Miller BB, Swerdlow RH, Dickson DW, Wszolek ZK. Mutations in the colony stimulating factor 1 receptor (CSF1R) cause hereditary diffuse leukoencephalopathy with spheroids. **Nature Genetics**. 2011. 44(2), 200-205. PMID: 22197934 PMCID: PMC3267847.
 - c. Fiskerstrand T, Arshad N, Haukanes BI, Tronstad RR, Pham KDC, Johansson S, Håvik B, Tønder SL, Levy SE, Brackman D, Boman H, Biswas KH, Apold J, Hovdenak N, Visweswariah SS, and Knappskog PM. Familial Diarrhea Syndrome Caused by an Activating GUCY2C Mutation. **New England Journal of Medicine**. 2012. 366(17), 1586-1595. PMID: 22436048.
 - d. Carlson J, Scott LJ, Locke AE, Flickinger M, Levy S, Myers RM, Boehnke M, Kang HM, Li JZ, Zöllner S. Extremely rare variants reveal patterns of germline mutation rate heterogeneity in humans. **bioRxiv**. 2017:108290.
 - e. Chao HT, Davids M, Burke E, Pappas JG, Rosenfeld JA, McCarty AJ, Davis T, Wolfe L, Toro C, Tift C, Xia F, Stong N, Johnson TK, Warr CG, Undiagnosed Diseases N, Yamamoto S, Adams DR, Markello TC, Gahl WA, Bellen HJ, Wangler MF, Malicdan MC. A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. **Am J**

Hum Genet. 2017;100(1):128-37. doi: 10.1016/j.ajhg.2016.11.018. PubMed PMID: 28017372; PMCID: PMC5223093.

6. The COVID pandemic presented unique opportunities to apply the genomic technologies and resources of my laboratory and collaborate with researchers from around the world to support the efforts to better understand and treat COVID and its related conditions.
 - a. MacKay MJ, Hooker AC, Afshinnkoo E, Salit M, Kelly J, Feldstein JV, Haft N, Schenkel D, Nambi S, Cai Y, Zhang F, Church G, Dai J, Wang CL, **Levy S**, Huber J, Ji HP, Kriegel A, Wyllie AL, and Mason, CE. (2020) *The COVID-19 XPRIZE and the need for scalable, fast, and widespread testing*. Nat Biotechnol. 38(9): 1021-1024, doi: 10.1038/s41587-020-0655-4, PMID: 32820257, PMC: PMC7543743.
 - b. Butler D, Mozsary C, Meydan C, Foox J, Rosiene J, Shaiber A, Danko D, Afshinnkoo E, MacKay M, Sedlazeck FJ, Ivanov NA, Sierra M, Pohl D, Zietz, M, Gisladdottir U, Ramlall V, Sholle ET, Schenck EJ, Westover CD, Hassan C, Ryon K, Young B, Bhattacharya C, Ng DL, Granados AC, Santos YA, Servellita V, Federman S, Ruggiero P, Fungtammasan A, Chin CS, Pearson NM, Langhorst BW, Tanner NA, Kim Y, Reeves JW, Hether TD, Warren SE, Bailey M, Gawrys J, Meleshko D, Xu D, Couto-Rodriguez M, Nagy-Szakal D, Barrows J, Wells H, O'Hara NB, Rosenfeld JA, Chen Y, Steel PAD, Shemesh AJ, Xiang J, Thierry-Mieg J, Thierry-Mieg D, Iftner A, Bezdan D, Sanchez E, Champion TR, Jr., Siple J, Cong L, Craney A, Velu P, Melnick AM, Shapira S, Hajirasouliha I, Borczuk A, Iftner T, Salvatore M, Loda M, Westblade LF, Cushing M, Wu S, **Levy S**, Chiu C, Schwartz RE, Tatonetti N, Rennert H, Imielinski M, and Mason CE. (2021) Shotgun transcriptome, spatial omics, and isothermal profiling of SARS-CoV-2 infection reveals unique host responses, viral diversification, and drug interactions. Nat Commun. 12(1): 1660, doi: 10.1038/s41467-021-21361-7, PMID: 33712587, PMC: PMC7954844.
 - c. Park J, Foox J, Hether T, Danko D, Warren S, Kim Y, Reeves J, Butler DJ, Mozsary C, Rosiene J, Shaiber A, Afshinnkoo E, MacKay M, Bram Y, Chandar V, Geiger H, Craney A, Velu P, Melnick AM, Hajirasouliha I, Beheshti A, Taylor D, Saravia-Butler A, Singh U, Wurtele ES, Schisler J, Fennessey S, Corvelo A, Zody MC, Germer S, Salvatore S, **Levy S**, Wu S, Tatonetti N, Shapira S, Salvatore M, Loda M, Westblade LF, Cushing M, Rennert H, Kriegel AJ, Elemento O, Imielinski M, Borczuk AC, Meydan C, Schwartz RE, and Mason CE. (2021) Systemic Tissue and Cellular Disruption from SARS-CoV-2 Infection revealed in COVID-19 Autopsies and Spatial Omics Tissue Maps. bioRxiv. doi: 10.1101/2021.03.08/434433, PMID: 33758858, PMC: PMC7987017.
 - d. Karlebach G, Aronow B, Baylin SB, Butler D, Foox J, **Levy S**, Meydan C, Mozsary C, Saravia-Butler AM, Taylor DM, Wurtele E, Mason CE, Beheshti A, and Robinson PN. (2021) Betacoronavirus-specific alternate splicing. bioRxiv. doi: 10.1101/2021.07.02.450920, PMID: 34230929, PMC: PMC8259905.

Education

College

University of New Hampshire: BS, 1994 (Biochemistry, Microbiology)
GPA 3.37
Honors Graduate, Dean's list.

Graduate School

Emory University: PhD, 2000, (Biochemistry)
GPA 3.75

Thesis title: "Genetic Alteration of the Mouse Mitochondrial Genome and Effects on Gene Expression."

Thesis advisor: Professor Douglas C. Wallace

Post-Graduate Training

Emory University, Douglas C. Wallace, March 2000-July 2000

Employment/Academic Appointments

Research Assistant Professor, Department of Molecular Physiology and Biophysics, Vanderbilt University Medical Center, Nashville, TN, July 2000-June 2003

Adjunct Faculty, Graduate training program, Department of Biomedical Informatics, Vanderbilt University Medical Center, Nashville, TN, January 2001-June 2003

Director, Vanderbilt Microarray Shared Resource, Vanderbilt University Medical Center, Nashville, TN, July 2000-August 2009

Assistant Professor, Department of Biomedical Informatics, Vanderbilt University Medical Center, Nashville, TN, July 2003-August 2009. (*Primary Appointment*)

Assistant Professor, Department of Molecular Physiology and Biophysics, Vanderbilt University Medical Center, Nashville, TN, July 2003-August 2009 (*Secondary Appointment*)

Adjunct Associate Professor, Department of Biomedical Informatics, Vanderbilt University Medical Center, Nashville, TN, August 2009-December 2017.

Adjunct Associate Professor, Department of Epidemiology, University of Alabama-Birmingham, Birmingham, AL October 2010-Present.

Adjunct Assistant Professor, Department of Genetics, University of Alabama-Birmingham, Birmingham, AL October 2010-Present.

Adjunct Associate Professor, Department of Biological Sciences, University of Alabama-Huntsville, Huntsville, AL January 2014-Present.

Director, Genomic Services Laboratory, HudsonAlpha Institute for Biotechnology, Huntsville, AL, August 2009-August 2019

Executive Director, HudsonAlpha Clinical Services Laboratory, LLC, Huntsville, AL, December 2014-August 2019

Faculty Investigator, HudsonAlpha Institute for Biotechnology, Huntsville, AL, August 2009-December 2022.

Chief Scientific Officer, HudsonAlpha Discovery, Discovery Life Sciences, Inc, Huntsville, AL, August 2019-March 2022.

Senior Vice President, Applications and Scientific Affairs, Element Biosciences, San Diego CA, February 2022-January 2023.

Chief Scientific Officer, Senior Vice President, Applications, Element Biosciences, San Diego CA, January 2023-present.

Professional Organizations

American Medical Informatics Association, Co-chair, Genomics Working Group (2006-2007)
Association of Biomedical Resource Facilities
American Association for the Advancement of Science
American Association for Cancer Research
American Society for Human Genetics

Professional Activities

Intramural-University

Vision 2020 Personalized Medicine Committee-Task Force 3 (2009)

Intramural-Departmental

Department of Biomedical Informatics Academic Progress Committee (2005-2007)
Department of Biomedical Informatics Curriculum Committee (2007-2009)

Intramural-Center Affiliations

Vanderbilt-Ingram Cancer Center, Associate Member (2000-2009)
Vanderbilt Diabetes Research and Training Center, Member (2000-2009)
Vanderbilt Digestive Disease Research Center, Member (2003-2009)
Vanderbilt Institute of Chemical Biology, Member (2004-2009)

Extramural-Journal Review

- Reviewer- Arteriosclerosis, Thrombosis and Vascular Biology (2001-present)
- Reviewer-Bioinformatics (2001-present)
- Reviewer-Journal of Biological Chemistry (2002-present)
- Reviewer-Neuropsychopharmacology (2003-present)
- Reviewer-Kidney International (2003-present)
- Reviewer-Circulation Research (2003-present)
- Reviewer-Proceedings of the National Academy of Sciences (2004-present)
- Reviewer-Mitochondrion (2004-present)
- Reviewer-Molecular Nutrition and Food Research (2005-present)
- Reviewer-Pattern Recognition Letters (2006-present)
- Reviewer-PLOS-Genetics (2006-present)
- Reviewer-Physiological Genomics (2008-present)
- Reviewer-Genome Biology (2008-present)

Extramural-Editorial

- Member, Editorial Board- Journal of the American Informatics Association (2005-2007)

Extramural-Grant Study Section

- Reviewer- Alzheimer's Association (2002-present).
- NIDDK study section ZDK1 GRB-6 "Digestive Disease Research Development Centers" December 2002.
- NIDDK study section ZDK1 GRB-6 "Digestive Disease Research Development Centers" April 2004.
- NCI study section ZCA1 SRRB-C "Innovative Technologies for the Detection of Cancer" July 2004.
- NLM special study section-P41 Biomedical Informatics Resource Grants, April 2005.
- NLM special emphasis panel ZLM1 HS RO1, July 2005

- NIH CSR shared equipment study section ZRG1 GGG-T (30, 31), November 2005.
- DOD Ovarian Cancer Review Panel OC-2, August 2006
- NIH Special Emphasis Panel ZRG1 GGG-T Genomics and Genetics Shared Instrumentation, October 2006.
- NCI study section ZCA1 SRRB-U Development of Advanced Genomic Characterization Technologies, November 2006.
- NIDDK DK-06-017 “Silvio O. Conte Digestive Diseases Research Core Centers P30”, June 2007.
- NIH Special Emphasis Panel ZRG1 GGG-A (30) - S10s genomics and proteomics shared instrumentation, July 2007.
- NIH Special Emphasis Panel ZRG1 GGG-B (30) - S10s genomics and proteomics shared instrumentation, September 2008.
- NIAAA Special Review Panel ZAA1-GG-01, November 2008
- NIH Special Emphasis Panel ZRG1 GGG-A (30) – Genes Genomes and Genetics instrumentation, October 2010.
- NIH Study Section 2011/05 GHD-Genetics of Health and Disease Study Section, February 2011.
- NIGRI Study Section 2012/05 ZHG1 HGR-P (M1) 1-H3 AFRICA Initiative, March 2012.

▪**Numerous ad-hoc reviews from 2012-2021.**

Extramural-Other Review

- Reviewer, American Association for the Advancement of Science Research Competitive Service-*Microarray Facilities for the Vermont Genetics Network*. April 2002.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Michigan Core Technology Alliance*. April 2003.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Michigan Core Technology Alliance*. April 2004.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Rhode Island EPScOR*. January 2007.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Rhode Island EPScOR*. March 2008.
- Reviewer, American Association for the Advancement of Science Research Competitive Service- *Review of Washington State Life Sciences Discovery Fund* June 2008.
- Reviewer, American Association for the Advancement of Science Research Competitive Service- *Review of Missouri Life Sciences Research Board* October 2008
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Rhode Island EPScOR*. June 2009.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Rhode Island EPScOR*. May 2010.
- Reviewer, American Association for the Advancement of Science Research Competitive Service-*External Review of the Rhode Island EPScOR*. September 2011.

Extramural-Advisory

- Member, Scientific Advisory Board, NuGen Technologies, Inc, San Carlos, CA, October 2003-December 2010.
- Member, Scientific Advisory Board, Genome Quebec Innovation Centre, Montreal, Quebec, 2008-2011.
- Member, Scientific Advisory Board, Rubicon Genomics, Ann Arbor, MI 2013-2017.

- Chairman, Scientific Advisory Board, RainDance Technologies (BioRad), Billerica, MA 2015-2018
- Member, Scientific Advisory Board, Genome Explorations Inc, Memphis, TN, 2006-2018.
- Member, Scientific Advisory Board, Jumpcode Genomics, San Diego, CA. 2018-Present.
- Member, Scientific Advisory Board, Covaris, Woburn, MA. 2018-Present.

Honors and Awards

- Scholar Athlete, University of New Hampshire, 1993-1994.
- Dean's list, University of New Hampshire, 1992-1994.
- Career Development Award, SPORE in Gastrointestinal Cancer 2004-2005
- Co-Chair, Genomics Working Group of the American Medical Informatics Association 2006-2007.

Teaching Activities

Graduate School Courses as Course Director

BMIF 310-Foundations of Bioinformatics and Computational Biology, 28 lectures, Spring 2004

BMIF 311-Introduction to Systems Biology, 28 lectures, Spring 2009. *This course was a newly developed course for 2009.*

Graduate School Courses as Lecturer

MPB 322-Regulation of Gene Expression, 3 lectures, Spring 2002

MPB 322-Regulation of Gene Expression, 2 lectures, Spring 2003

MPB 322-Regulation of Gene Expression, 3 lectures, Spring 2004

IGP 301-Methodology, 1 lecture, Fall 2004

IGP 301-Methodology, 1 lecture, Fall 2005

IGP 301-Methodology, 1 lecture, Fall 2006

MIM 351-Functional Genomics and Proteomics, 2 lectures, Spring 2006

BMIF 310-Foundations of Bioinformatics and Computational Biology, 7 lectures, Fall 2007

BMIF 310-Foundations of Bioinformatics and Computational Biology, 7 lectures, Fall 2008

BMIF 310-Foundations of Bioinformatics and Computational Biology, 4 lectures, Fall 2009

BMIF 310-Foundations of Bioinformatics and Computational Biology, 4 lectures, Fall 2010

BMIF 310-Foundations of Bioinformatics and Computational Biology, 1 lecture, Fall 2011

Research Supervision

Ph.D. Thesis Committee Member

Stephen VonStetina-Vanderbilt University (2001-2005)

Laura Wilding-Vanderbilt University (2003-2007)

Alex Statnikov-Vanderbilt University (2005-2008)

Alisha Russell-Vanderbilt University (2006-2010)

Mawuli Nyaku-University of Alabama-Birmingham (2010-2014)

M.S. Thesis Committee Member

Alex Statnikov (2003-2005)

Joel Parker (2000-2002)

Student Mentorship

Shristi Shrestha, PhD student (2014-2019)

Current position: Bioinformatics Scientist, Department of Stem Cell Biology, Vanderbilt University, Nashville, TN.

Nripesh Prasad, PhD student (2010-2014)

Current position: Director of Feasibility, Discovery Life Sciences, Huntsville, AL.

Sidd Pratrapp MS student (2005-2007)

Current position: Director of Bioinformatics, Meharry Medical College, Nashville, TN.

Fellow Mentorship

Lewis Frey, PhD (2004-2006)

Current position: Associate Professor, Public Health Sciences, Medical College of South Carolina, Charleston, SC.

Patents Awarded

Multiplex spatial profiling of gene expression
US 7,569,392 B2

Research Support

COMPLETED

2U24HD090744-01 (Levy/Zhang)	07/01/2019 – 06/30/2022	2.40 calendar months
NIH/NICHD	\$6,212,400	

Characterizing pediatric genomes through an optimized sequencing approach

Understanding the fundamental genetic changes associated with structural birth defects and childhood cancers is an important step in developing tools to allow more advanced prediction, treatment and prevention of these devastating conditions. We propose to combine the resources of two world-class centers to support researchers in their investigations of the genetics of birth defects and childhood cancers. This centralized resource will provide researchers with the tools and support necessary to advance our understanding and drive us closer to curing or preventing these diseases.

Role: Principle Investigator

1OT2OD027070-01 (Levy)	09/21/2019 - 08/31/2023	2.40 calendar months
NIH/NCATS	\$6,999,491	

Population-Scale Clinical Genomics

The production of genomic data for the All of Us Research Program (AoU) is an unprecedented opportunity to support a comprehensive precision medicine effort that benefits the entire U.S. population. With the participation from patients, family members, researchers and clinicians, the AoU Research Program will impact clinical management by directing patient care, enabling pre-symptomatic genetic testing of relatives, guiding family planning measures, and triggering healthcare interventions. This proposal focuses on the production of long-read sequencing data to complement the short-read data that is being produced from DNA from participating individuals. The goal of the study is to identify structural and copy number changes detected in the long-read data that is may be missed in short-read data.

Role: Principle Investigator

5 U24 DK58749-03 (George)	09/30/2000 - 08/31/2003	1.2 calendar months
NIH/NIDDK		

Vanderbilt NIDDK Biotechnology Center

The goal of this proposal was the establishment of a Biotechnology Center for the support of genomic studies of interest to investigators funded by the NIDDK. Microarray technologies and related informatics were central to the efforts.

Role: Co-Investigator

VUMC Discovery Grant 540 (Levy)

01/01/2002 - 12/31/2003

1.2 calendar months

VUMC Internal Grant

\$50,000

Gene Expression Analysis of Colon Cancer

The goal of this proposal was the development of an integrated RNA and protein expression profile for colon cancer utilizing microarray and high-resolution protein profiling technologies. These profiles were useful in designing and developing both technological and informatic platforms for the combined analysis of protein and genetic profiles of cancer.

Role: Principle Investigator

ACS IRG-58-009-46 (Levy)

07/01/2003 - 06/30/2004

1.2 calendar months

ACS/VICC

Simultaneous profiling of protein and RNA expression by mass spectrometry in intact tissue sections.

The goal of this proposal is to develop a novel technology platform that facilitates the simultaneous profiling of protein and RNA species in intact tissue samples while reporting spatial position. This will provide an unprecedented resolution to examine the biology of tumor samples and host-tumor interactions.

Role: Principle Investigator

1 R21 NS043581-01A1 (McDonald)

12/01/2002 - 11/30/2004

1.2 calendar months

NIH/NINDS

Gene Discovery in a Putative Mouse Model of ADHD

In this proposal, microarray technology will be used to examine differential gene expression in the mouse model of ADHD, providing a rare opportunity to discover genes downstream of TR β activity that are able to produce all of the core symptoms and many adjunct features of ADHD.

Role: Co-Investigator

1 U01 DK063587-01 (Hayward)

09/30/2002 - 06/30/2005

1.2 calendar months

NIH/NIDDK

Genetic Markers of Transition Zone Hyperplasia

The goals of this proposal are the identification of biomarkers for prostate hyperplasia through the use of high-density microarray studies on novel models of prostate disease.

Role: Co-Investigator

W81XWH-04-1-0626 (Levy)

07/15/2004 - 07/14/2006

1.2 calendar months

Department of Defense

Simultaneous profiling of protein and RNA expression by mass spectrometry in intact breast tissue sections.

The goal of this proposal is to continue the development of a novel technology platform that facilitates the simultaneous profiling of protein and RNA species in intact tissue samples while reporting spatial position. This proposal will specifically fund the optimization of this technology for the analysis of breast tissue samples.

Role: Principle Investigator

5 P01 HL6744-04 (Hawiger)

12/01/2001 - 11/30/2006

1.2 calendar months

NIH/NHLBI

Functional Genomics of Inflammation

As part of a Program Project Grant, the goal of the Microarray Core in the Functional Genomics of Inflammation program project is to provide genome-scale expression profiling technologies to researchers involved in the program.

Role: Core Leader**1 R01 DK068261-01 (Nagy)**

07/01/2004 - 06/30/2007

1.2 calendar months

NIH/NIDDK

Antipsychotic Drug-induced Weight Gain

The goal of this study is to understand the actions of antipsychotic drugs as they alter body weight. In this short subcontract with the University of Alabama, an animal model system used to study the molecular effects of selected drugs will be analyzed using genomic profiling techniques.

Role: Co-Investigator**5 P60 DK20593-27 (Powers)**

07/20/2002 - 03/31/2007

1.2 calendar months

NIH/NIDDK

Diabetes Research and Training Center-Microarray and Bioinformatics Core

As part of a center grant, the goal of the Microarray Core in the Diabetes Research and Training Center is to provide support for the use of genome-scale expression profiling technologies to researchers involved in DRTC-related research.

Role: Core Leader**5 P50 CA95103-04 (Coffey)**

09/24/2002 - 04/30/2007

1.2 calendar months

NIH/NCI

SPORE in GI Cancer

This study will investigate the molecular features of tumors in GI cancer and provide full support for genomic profiling projects as part of the overall SPORE program.

Role: Core Leader**5 P30 CA68485-13 (Pietenpol)**

09/28/2004 - 08/31/2009

1.80 calendar months

NIH/NCI

\$3,553,801

Cancer Center Support Grant

As part of the Vanderbilt Ingram Cancer Center's support grant, the goal of the Microarray Core is to provide genome-scale expression profiling technologies as well as analysis and informatics support to researchers who are members of the center.

Role: Core Leader**U24 CA126563 (Myers)**

09/28/2006 - 08/31/2010

1.2 calendar months

NIH/NCI

The HudsonAlpha Cancer Genome Characterization Center

We are characterizing tumors and matched non-tumor samples for copy number variations throughout the human genome as part of The Cancer Genome Atlas project, a trans-NIH initiative aimed at learning all the genetic and genomic changes associated with cancer. We use a whole-genome genotyping method to assay more than 1 million SNPs throughout the genome.

Role: Co-Investigator**1 RC1 HL100016-01 (Schey)**

09/30/2009 - 09/29/2011

1.2 calendar months

NIH-ARRA Funding

Proteome and Transcriptome Markers of Hypertension in Urine and Plasma Exosomes

The goal of the proposed research is to develop a novel method for discovery of molecular markers of disease that circumvents existing obstacles. Through analysis of proteins and RNA found in lipid particles isolated from blood and urine, new markers of disease will be discovered that improve diagnosis, prognosis, and prediction of response to therapy; that is, improve personalized medicine. The new methodology will be applied to reveal biomarkers of salt-sensitivity and therapeutic response in hypertensive subjects.

Role: Co-Investigator

1 RC1 DK086594-01 (Southard-Smith) 09/30/2009 - 09/29/2011 0.60 calendar months
NIH/NIDDK \$240,970

Gene Networks in Neutral Crest-derived Innervation of the Lower Urinary Tract

The studies proposed aim to identify essential genes that control development of nerves in the lower urinary tract that regulate bladder control and sexual function. These studies are important for understanding how these nerves normally develop and for deriving technologies that will restore neural function in urogenital birth defects or after pelvic surgery. This proposal is in response to the broad Challenge grant area of Regenerative medicine and meets multiple needs for basic research in development lower urinary tract innervation.

Role: Co-Investigator

5 P30DK058404-07 (Polk) 08/30/2007 - 05/31/2012 1.20 calendar months
NIH/NIDDK \$727,500

Molecular and Cellular Basis of Digestive Diseases

As part of a center grant, the goal of the Microarray Core in the Vanderbilt Digestive Diseases Center is to provide support for the use of genome-scale expression profiling technologies to researchers involved in digestive disease-related research.

Role: Core Leader

5 P60 DK20593-31 (Powers) 06/01/2007 - 03/31/2012 0.24 calendar months
NIH/NIDDK \$1,487,659

Diabetes Research and Training Center

As part of a center grant, the goal of the Microarray and Bioinformatics Core in the Diabetes Research and Training Center is to provide support for the use of genome-scale expression profiling technologies to researchers involved in DRTC-related research.

Role: Core Leader

P50 HG02568 (Kingsley) 4/19/2002 - 5/31/2012 1.20 calendar months
NIH/NHGRI \$701,981

Center for Vertebrate Diversity

The continuation of this Center of Excellence in Genome Science (CEGS) has broad goals to understand the genetic basis for the striking biological diversity seen in vertebrate animals. We use genetics, genomics, molecular biology and computational tools to study this problem, focusing on the three-spined stickleback fish. HudsonAlpha performs many of the genomic experiments for this project, including genomic DNA sequencing, cDNA sequencing, BAC map construction, and genotyping.

Role: Co-Investigator

2 R01 CA064277-10A1 (Zheng) 08/05/2008 - 05/31/2013 0.24 calendar months
NIH/NCI \$324,917

Shanghai Breast Cancer Study

This proposal is aimed at the development of novel algorithms for the analysis of high-dimensionality data towards to the discovery of causal markers and mechanisms.

Role: Co-Investigator

R01: (Myers and Boehnke) 8/30/11 - 6/30/14 2.4 calendar months
NIH \$1,855,348

Whole Genome and Exome Sequencing for Bipolar Disorder

In this collaborative R01 grant, performed jointly with Dr. Michael Boehnke and colleagues at the University of Michigan, we are performing a detailed genetic analysis of bipolar disorder. We are using ultrahigh-throughput sequencing to determine the deep whole genome sequences from 1,000 individuals with bipolar disorder and 1,000 control individuals without the disorder.

Role: Co-Investigator

1 R01 AR057202 (Bridges) 4/1/2009 - 3/31/2014 0.6 calendar months
NIH/NIAMS \$298,704

Genome Wide Association Study in African-Americans with Rheumatoid Arthritis

In this study, the Myers lab and Devin Absher and his lab at HudsonAlpha are collaborating with Dr. Lou Bridges and his colleagues at the School of Medicine at the University of Alabama in Birmingham to perform a genome-wide genetic association study of rheumatoid arthritis in African Americans. No budgetary or scientific overlap.

Role: Co-Investigator

US MED Research ACQ Activity (Myers) 9/16/2010 - 8/31/2015 4.0 calendar months
US Army \$2,150,777

Global genomic analysis of prostate, breast and pancreatic cancer

The goals of this study are to provide an unprecedented comprehensive view of the molecular pathogenesis of prostate, breast, and pancreatic cancer, as well as the differential response to treatments in breast cancer. We will use next-generation DNA sequencing to measure mRNA, microRNA, DNA methylation, DNase hypersensitivity sites, histone modifications, and sites of transcription factor occupancy in tumors and matched non-tumor tissues for these three cancers.

Role: Co-Investigator

5 U54 HG004576-03 (Myers) 10/01/2007 - 09/30/2011 1.20 calendar months
NIH/NHGRI \$3,985,643

Global Annotation of Regulatory Elements in the Human Genome

This project, which is a collaboration between the Myers group at HudsonAlpha and Barbara Wold's group at Caltech, along with contributions from Wing Wong, Arend Sidow, Serafim Batzoglou and Gavin Sherlock at Stanford, is part of the ENCODE Project, whose goals are to identify and understand the roles of all the functional elements throughout the entire human genome. Our contributions are to identify transcription factor binding sites, assess the methylation status and measure RNAs with next-gen sequencing.

Role: Co-Investigator

5UL1TR001417-02 (Kimberly) 08/18/2015 - 03/31/2019 0.60 calendar months
NIH/NCATS \$83,644

UAB Center for Clinical and Translational Science (CCTS)

The UAB CCTS will enhance human health by driving scientific discovery and dialogue across the bench, bedside and community continuum. The CCTS support this overall mission in a highly integrative network of relationships. Success in creating such an environment is dependent upon success in achieving five strategic priorities: 1) enhancing research infrastructure; 2) promoting

investigator education, training and development; 3) accelerating discovery across the T1 interface; 4) expanding value-added partnerships; and 5) building sustainability.

Role: Co-Investigator

HHSN2722012000231 (Creech)

09/01/2015 - 09/30/2018

0.24 calendar months

NIH/NIAID

\$555,660

Influenza A/H7N9 Vaccine Administered with/without AS03 Adjuvant: Standard and Systems Biology

HudsonAlpha will receive human RNA samples from Vanderbilt University Medical Center. RNA-sequencing will be performed per specifications provided in the clinical protocol and clarified in the manual of procedures. We will perform all necessary experiments, including quality control assays. Once sequencing data are obtained, these FastQ/BAM files will be transferred to Vanderbilt University Medical Center and to the DMID Statistics and Data Coordinating Center (SDCC) for data analysis.

Role: Co-Investigator

HHSN2722012000231 (Creech)

09/01/2015 - 09/30/2018

0.24 calendar months

NIH/NIAID

\$56,630

Sub-study for DMID 10-0074

HudsonAlpha will receive human RNA samples from Vanderbilt University Medical Center. RNA-sequencing will be performed per specifications provided in the clinical protocol and clarified in the manual of procedures. We will perform all necessary experiments, including quality control assays. Once sequencing data are obtained, these FastQ/BAM files will be transferred to Vanderbilt University Medical Center and to the DMID Statistics and Data Coordinating Center (SDCC) for data analysis.

Role: Co-Investigator

6U19CA179514-05 (Coffey)

09/01/2013 - 08/31/2018

0.24 calendar months

NIH/NCI

\$39,254

Secreted RNA during CRC progression biogenesis function and clinical markers

Dr. Levy's laboratory will fully support RNA sequencing on 48-74 samples per year prepared from either total RNA or microRNA at the HudsonAlpha Institute for Biotechnology. Dr. Levy's laboratory will provide all required reagents, personnel and basic analysis support for the proposed sequencing studies during years 1-5 of the project period.

Role: Co-Investigator

5U01MH105653-03 (Boehnke)

09/19/2014 - 05/31/2018

0.60 calendar months

NIH/NIMH

\$23,557

Whole Genome Sequencing for Schizophrenia and Bipolar Disorder in the GPC

Dr. Levy will participate in weekly conference calls and several yearly face-to-face meetings to help make this project successful. Any new improvements in sequencing technology, data analysis and data interpretation that are developed and/or applied at HudsonAlpha will be made immediately available to this project.

Role: Co-Investigator

3P30CA013145-44S4 (Partridge)

04/01/2017 - 03/31/2018

0.60 calendar months

NIH/NCI

\$113,863

Comprehensive Cancer Center Core Support Grant

Dr. Myers, President and Science Director of HudsonAlpha Institute for Biotechnology, will be part of the director's council. The director's council meets on a monthly basis to advise the director on all major decisions regarding the UAB-CCC, its organization, planning and evaluation and to

approve new developmental research programs and review program leaderships. In addition, Dr. Myers will co-lead UAB-CCC's Experimental Therapeutics program. Drs. Absher and Levy will be co-leaders of the Cancer Cell Biology Program and Cancer Control & Population Sciences Program. Dr. Cooper is an Associate Scientist in Experimental Therapeutics program. They will consult investigators in study design and analysis related to genomic data.

Role: Core Leader

4UM1HG007301-04 (Cooper/Myers) 06/14/2013 - 05/31/2018 0.60 calendar months
NIH/NHGRI \$1,536,927

Genomic Diagnosis in Children with Developmental Delay

The goal of this project is to address technological, analytical, and ethical challenges that prevent optimal use of DNA sequencing to improve treatment of diseases and life planning for patients and their families. We are applying next-generation DNA sequencing to meet the diagnostic needs of children with developmental delay, intellectual disability and related health problems.

Role: Co-Investigator

RFA-HG-16-011 (Cooper/Barsh/Korf) 06/01/2017 – 05/31/2021 0.60 calendar months
NIH \$2,840,944

Clinical sequencing across communities in the Deep South

This proposal outlines an important study to apply WGS to diagnose neonates with rare disorders, increase participation of individuals from underrepresented racial/ethnic groups in genomics clinical trials, provide educational materials appropriate to diverse audiences, equip non-genetics healthcare providers to return WGS results, assess the impact of WGS testing and results, and engage a broad community to implement safer, more effective, and more equitably distributed genomic medicine.

Role: Co-Investigator

Publications

221 peer-reviewed publications with a total of 55,665 citations (as of November 2023).

A full publication and patent listing can be accessed via a public Google Scholar profile at: <http://scholar.google.com/citations?user=xeKJAZ0AAAAJ>

NCBI Bibliography Link:

<https://www.ncbi.nlm.nih.gov/myncbi/1BODvQqGn4iAa/bibliography/public/>

Articles in refereed journals

1. Levy, S.E., K.G. Waymire, Y.L. Kim, G.R. MacGregor, and D.C. Wallace, *Transfer of chloramphenicol-resistant mitochondrial DNA into the chimeric mouse*. Transgenic Res, 1999. **8**(2): p. 137-45.
2. Levy, S.E., Y.S. Chen, B.H. Graham, and D.C. Wallace, *Expression and sequence analysis of the mouse adenine nucleotide translocase 1 and 2 genes*. Gene, 2000. **254**(1-2): p. 57-66.
3. Sligh, J.E., S.E. Levy, K.G. Waymire, P. Allard, D.L. Dillehay, S. Nusinowitz, J.R. Heckenlively, G.R. MacGregor, and D.C. Wallace, *Maternal germ-line transmission of*

- mutant mtDNAs from embryonic stem cell-derived chimeric mice. Proc Natl Acad Sci U S A*, 2000. **97**(26): p. 14461-6.
4. Levy, S.E. and J.A. Muldowney, 3rd, *Microarray analysis of neointima: flowing toward a clear future. Arterioscler Thromb Vasc Biol*, 2002. **22**(12): p. 1946-7.
5. Gu, G., A.Y. Deutch, J. Franklin, S. Levy, D.C. Wallace, and J. Zhang, *Profiling genes related to mitochondrial function in mice treated with N-methyl-4-phenyl-1,2,3,6-tetrahydropyridine. Biochem Biophys Res Commun*, 2003. **308**(1): p. 197-205.
6. Levy, S.E., *Microarray analysis in drug discovery: an uplifting view of depression. Sci STKE*, 2003. **2003**(206): p. pe46.
7. McQuain, M.K., K. Seale, J. Peek, S. Levy, and F.R. Haselton, *Effects of relative humidity and buffer additives on the contact printing of microarrays by quill pins. Anal Biochem*, 2003. **320**(2): p. 281-91.
8. Yamagata, N., Y. Shyr, K. Yanagisawa, M. Edgerton, T.P. Dang, A. Gonzalez, S. Nadaf, P. Larsen, J.R. Roberts, J.C. Nesbitt, R. Jensen, S. Levy, J.H. Moore, J.D. Minna, and D.P. Carbone, *A training-testing approach to the molecular classification of resected non-small cell lung cancer. Clin Cancer Res*, 2003. **9**(13): p. 4695-704.
9. Friedman, D.B., S. Hill, J.W. Keller, N.B. Merchant, S.E. Levy, R.J. Coffey, and R.M. Caprioli, *Proteome analysis of human colon cancer by two-dimensional difference gel electrophoresis and mass spectrometry. Proteomics*, 2004. **4**(3): p. 793-811.
10. Kokoszka, J.E., K.G. Waymire, S.E. Levy, J.E. Sligh, J. Cai, D.P. Jones, G.R. MacGregor, and D.C. Wallace, *The ADP/ATP translocator is not essential for the mitochondrial permeability transition pore. Nature*, 2004. **427**(6973): p. 461-5.
11. Law, A.K., D. Gupta, S. Levy, D.C. Wallace, R.J. McKeon, and C.R. Buck, *TGF-beta1 induction of the adenine nucleotide translocator 1 in astrocytes occurs through Smads and Sp1 transcription factors. BMC Neurosci*, 2004. **5**: p. 1.
12. McQuain, M.K., K. Seale, J. Peek, T.S. Fisher, S. Levy, M.A. Stremler, and F.R. Haselton, *Chaotic mixer improves microarray hybridization. Anal Biochem*, 2004. **325**(2): p. 215-26.
13. Sforza, D.M., J. Annese, D. Liu, S. Levy, A.W. Toga, and D.J. Smith, *Anatomical methods for voxelation of the mammalian brain. Neurochem Res*, 2004. **29**(6): p. 1299-306.
14. Levy, S.E., A. Statnikov, and C. Aliferis, *Biomarker Selection from High-Dimensionality Data. Microarray Technol.(Suppl. Pharmaceut. Discov.)*, 2005: p. 37-44.
15. Park, Y.K., J.L. Franklin, S.H. Settle, S.E. Levy, E. Chung, L.H. Jeyakumar, Y. Shyr, M.K. Washington, R.H. Whitehead, B.J. Aronow, and R.J. Coffey, *Gene expression profile analysis of mouse colon embryonic development. Genesis*, 2005. **41**(1): p. 1-12.
16. Statnikov, A., C.F. Aliferis, I. Tsamardinos, D. Hardin, and S. Levy, *A comprehensive evaluation of multicategory classification methods for microarray gene expression cancer diagnosis. Bioinformatics*, 2005. **21**(5): p. 631-43.
17. Xie, L., B.J. Xu, A.E. Gorska, Y. Shyr, S.A. Schwartz, N. Cheng, S. Levy, B. Bieri, R.M. Caprioli, and H.L. Moses, *Genomic and proteomic analysis of mammary tumors arising in transgenic mice. J Proteome Res*, 2005. **4**(6): p. 2088-98.
18. Acuff, H.B., M. Sinnamon, B. Fingleton, B. Boone, S.E. Levy, X. Chen, A. Pozzi, D.P. Carbone, D.R. Schwartz, K. Moin, B.F. Sloane, and L.M. Matrisian, *Analysis of host- and tumor-derived proteinases using a custom dual species microarray reveals a protective role for stromal matrix metalloproteinase-12 in non-small cell lung cancer. Cancer Res*, 2006. **66**(16): p. 7968-75.
19. Chung, C.H., K. Ely, L. McGavran, M. Varella-Garcia, J. Parker, N. Parker, C. Jarrett, J. Carter, B.A. Murphy, J. Netterville, B.B. Burkey, R. Sinard, A. Cmelak, S. Levy, W.G. Yarbrough, R.J. Slebos, and F.R. Hirsch, *Increased epidermal growth factor receptor gene copy number is associated with poor prognosis in head and neck squamous cell carcinomas. J Clin Oncol*, 2006. **24**(25): p. 4170-6.

20. Chung, C.H., S. Levy, and W.G. Yarbrough, *Clinical applications of genomics in head and neck cancer*. Head Neck, 2006. **28**(4): p. 360-8.
21. Chung, C.H., J.S. Parker, K. Ely, J. Carter, Y. Yi, B.A. Murphy, K.K. Ang, A.K. El-Naggar, A.M. Zanation, A.J. Cmelak, S. Levy, R.J. Slebos, and W.G. Yarbrough, *Gene expression profiles identify epithelial-to-mesenchymal transition and activation of nuclear factor-kappaB signaling as characteristics of a high-risk head and neck squamous cell carcinoma*. Cancer Res, 2006. **66**(16): p. 8210-8.
22. Shi, L., L.H. Reid, W.D. Jones, R. Shippy, J.A. Warrington, S.C. Baker, P.J. Collins, F. de Longueville, E.S. Kawasaki, K.Y. Lee, Y. Luo, Y.A. Sun, J.C. Willey, R.A. Setterquist, G.M. Fischer, W. Tong, Y.P. Dragan, D.J. Dix, F.W. Frueh, F.M. Goodsaid, D. Herman, R.V. Jensen, C.D. Johnson, E.K. Lobenhofer, R.K. Puri, U. Schrf, J. Thierry-Mieg, C. Wang, M. Wilson, P.K. Wolber, L. Zhang, S. Amur, W. Bao, C.C. Barbacioru, A.B. Lucas, V. Bertholet, C. Boysen, B. Bromley, D. Brown, A. Brunner, R. Canales, X.M. Cao, T.A. Cebula, J.J. Chen, J. Cheng, T.M. Chu, E. Chudin, J. Corson, J.C. Corton, L.J. Croner, C. Davies, T.S. Davison, G. Delenstarr, X. Deng, D. Dorris, A.C. Eklund, X.H. Fan, H. Fang, S. Fulmer-Smentek, J.C. Fuscoe, K. Gallagher, W. Ge, L. Guo, X. Guo, J. Hager, P.K. Haje, J. Han, T. Han, H.C. Harbottle, S.C. Harris, E. Hatchwell, C.A. Hauser, S. Hester, H. Hong, P. Hurban, S.A. Jackson, H. Ji, C.R. Knight, W.P. Kuo, J.E. LeClerc, S. Levy, Q.Z. Li, C. Liu, Y. Liu, M.J. Lombardi, Y. Ma, S.R. Magnuson, B. Maqsodi, T. McDaniel, N. Mei, O. Myklebost, B. Ning, N. Novoradovskaya, M.S. Orr, T.W. Osborn, A. Papallo, T.A. Patterson, R.G. Perkins, E.H. Peters, R. Peterson, K.L. Philips, P.S. Pine, L. Pusztai, F. Qian, H. Ren, M. Rosen, B.A. Rosenzweig, R.R. Samaha, M. Schena, G.P. Schroth, S. Shchegrova, D.D. Smith, F. Staedtler, Z. Su, H. Sun, Z. Szallasi, Z. Tezak, D. Thierry-Mieg, K.L. Thompson, I. Tikhonova, Y. Turpaz, B. Vallanat, C. Van, S.J. Walker, S.J. Wang, Y. Wang, R. Wolfinger, A. Wong, J. Wu, C. Xiao, Q. Xie, J. Xu, W. Yang, L. Zhang, S. Zhong, Y. Zong and W. Slikker, Jr., *The MicroArray Quality Control (MAQC) project shows inter- and intraplatform reproducibility of gene expression measurements*. Nat Biotechnol, 2006. **24**(9): p. 1151-61.
23. Slebos, R.J., Y. Yi, K. Ely, J. Carter, A. Evjen, X. Zhang, Y. Shyr, B.M. Murphy, A.J. Cmelak, B.B. Burkey, J.L. Netterville, S. Levy, W.G. Yarbrough, and C.H. Chung, *Gene expression differences associated with human papillomavirus status in head and neck squamous cell carcinoma*. Clin Cancer Res, 2006. **12**(3 Pt 1): p. 701-9.
24. Chin, M.H., A.B. Geng, A.H. Khan, W.J. Qian, V.A. Petyuk, J. Boline, S. Levy, A.W. Toga, R.D. Smith, R.M. Leahy, and D.J. Smith, *A genome-scale map of expression for a mouse brain section obtained using voxelation*. Physiol Genomics, 2007. **30**(3): p. 313-21.
25. Chung, C.H., S. Levy, P. Chaurand, and D.P. Carbone, *Genomics and proteomics: emerging technologies in clinical cancer research*. Crit Rev Oncol Hematol, 2007. **61**(1): p. 1-25.
26. Chung, C.H., J. Parker, S. Levy, R.J. Slebos, A.P. Dicker, and U. Rodeck, *Gene expression profiles as markers of aggressive disease-EGFR as a factor*. Int J Radiat Oncol Biol Phys, 2007. **69**(2 Suppl): p. S102-5.
27. Frey, L., M. Edgerton, D. Fisher, and S. Levy, *Ensemble stump classifiers and gene expression signatures in lung cancer*. Stud Health Technol Inform, 2007. **129**(Pt 2): p. 1255-9.
28. Joshi, S., H. Davies, L.P. Sims, S.E. Levy, and J. Dean, *Ovarian gene expression in the absence of FIGLA, an oocyte-specific transcription factor*. BMC Dev Biol, 2007. **7**: p. 67.
29. Kaiser, S., Y.K. Park, J.L. Franklin, R.B. Halberg, M. Yu, W.J. Jessen, J. Freudenberg, X. Chen, K. Haigis, A.G. Jegga, S. Kong, B. Sakthivel, H. Xu, T. Reichling, M. Azhar, G.P. Boivin, R.B. Roberts, A.C. Bissahoyo, F. Gonzales, G.C. Bloom, S. Eschrich, S.L. Carter, J.E. Aronow, J. Kleimeyer, M. Kleimeyer, V. Ramaswamy, S.H. Settle, B. Boone, S. Levy, J.M. Graff, T. Doetschman, J. Groden, W.F. Dove, D.W. Threadgill, T.J. Yeatman, R.J.

- Coffey, Jr., and B.J. Aronow, *Transcriptional recapitulation and subversion of embryonic colon development by mouse colon tumor models and human colon cancer*. *Genome Biol*, 2007. **8**(7): p. R131.
30. Muldowney, J.A., 3rd, J.R. Stringham, S.E. Levy, L.A. Gleaves, M. Eren, R.N. Piana, and D.E. Vaughan, *Antiproliferative agents alter vascular plasminogen activator inhibitor-1 expression: a potential prothrombotic mechanism of drug-eluting stents*. *Arterioscler Thromb Vasc Biol*, 2007. **27**(2): p. 400-6.
31. Zhou, S., S. Wei, B. Boone, and S. Levy, *Microarray analysis of genes affected by salt stress in tomato*. *African Journal of Environmental Science and Technology*, 2007. **1**(2): p. 014-026.
32. Beeghly-Fadiel, A., J.R. Long, Y.T. Gao, C. Li, S. Qu, Q. Cai, Y. Zheng, Z.X. Ruan, S.E. Levy, S.L. Deming, J.R. Snoddy, X.O. Shu, W. Lu, and W. Zheng, *Common MMP-7 polymorphisms and breast cancer susceptibility: a multistage study of association and functionality*. *Cancer Res*, 2008. **68**(15): p. 6453-9.
33. Kanies, C.L., J.J. Smith, C. Kis, C. Schmidt, S. Levy, K.S. Khabar, J. Morrow, N. Deane, D.A. Dixon, and R.D. Beauchamp, *Oncogenic Ras and transforming growth factor-beta synergistically regulate AU-rich element-containing mRNAs during epithelial to mesenchymal transition*. *Mol Cancer Res*, 2008. **6**(7): p. 1124-36.
34. Liu, Y.J., X.G. Liu, L. Wang, C. Dina, H. Yan, J.F. Liu, S. Levy, C.J. Papasian, B.M. Drees, J.J. Hamilton, D. Meyre, J. Delplanque, Y.F. Pei, L. Zhang, R.R. Recker, P. Froguel, and H.W. Deng, *Genome-wide association scans identified CTNBL1 as a novel gene for obesity*. *Hum Mol Genet*, 2008. **17**(12): p. 1803-13.
35. Liu, Y.Z., S.G. Wilson, L. Wang, X.G. Liu, Y.F. Guo, J. Li, H. Yan, P. Deloukas, N. Soranzo, U. Chinappan-Horsley, A. Cervino, F.M. Williams, D.H. Xiong, Y.P. Zhang, T.B. Jin, S. Levy, C.J. Papasian, B.M. Drees, J.J. Hamilton, R.R. Recker, T.D. Spector, and H.W. Deng, *Identification of PLCL1 gene for hip bone size variation in females in a genome-wide association study*. *PLoS One*, 2008. **3**(9): p. e3160.
36. Subramaniam, V., P. Golik, D.G. Murdock, S. Levy, K.W. Kerstann, P.E. Coskun, G.A. Melkonian, and D.C. Wallace, *MITOCHIP assessment of differential gene expression in the skeletal muscle of Ant1 knockout mice: coordinate regulation of OXPHOS, antioxidant, and apoptotic genes*. *Biochim Biophys Acta*, 2008. **1777**(7-8): p. 666-75.
37. Watson, J.D., S. Wang, S.E. Von Stetina, W.C. Spencer, S. Levy, P.J. Dexheimer, N. Kurn, J.D. Heath, and D.M. Miller, 3rd, *Complementary RNA amplification methods enhance microarray identification of transcripts expressed in the C. elegans nervous system*. *BMC Genomics*, 2008. **9**: p. 84.
38. Wilding Crawford, L., E. Tweedie Ables, Y.A. Oh, B. Boone, S. Levy, and M. Gannon, *Gene expression profiling of a mouse model of pancreatic islet dysmorphogenesis*. *PLoS One*, 2008. **3**(2): p. e1611.
39. Xu, B., J.L. Roos, S. Levy, E.J. van Rensburg, J.A. Gogos, and M. Karayiorgou, *Strong association of de novo copy number mutations with sporadic schizophrenia*. *Nat Genet*, 2008. **40**(7): p. 880-5.
40. Zhou, S., R.J. Suave, B. Boone, and S. Levy, *Identification of Genes Associated with Aluminum Toxicity in Tomato Roots using cDNA Microarrays*. *Plant Stress*, 2008. **2**(2): p. 113-120.
41. De Moor, M.H., Y.J. Liu, D.I. Boomsma, J. Li, J.J. Hamilton, J.J. Hottenga, S. Levy, X.G. Liu, Y.F. Pei, D. Posthuma, R.R. Recker, P.F. Sullivan, L. Wang, G. Willemsen, H. Yan, E.J. De Geus, and H.W. Deng, *Genome-wide association study of exercise behavior in Dutch and American adults*. *Med Sci Sports Exerc*, 2009. **41**(10): p. 1887-95.
42. Lei, S.F., L.J. Tan, X.G. Liu, L. Wang, H. Yan, Y.F. Guo, Y.Z. Liu, D.H. Xiong, J. Li, T.L. Yang, X.D. Chen, Y. Guo, F.Y. Deng, Y.P. Zhang, X.Z. Zhu, S. Levy, C.J. Papasian, J.J. Hamilton, R.R. Recker, and H.W. Deng, *Genome-wide association study identifies two*

- novel loci containing FLNB and SBF2 genes underlying stature variation.* Hum Mol Genet, 2009. **18**(9): p. 1661-9.
43. Li, J., T. Yang, L. Wang, H. Yan, Y. Zhang, Y. Guo, F. Pan, Z. Zhang, Y. Peng, Q. Zhou, L. He, X. Zhu, H. Deng, S. Levy, C.J. Papasian, B.M. Drees, J.J. Hamilton, R.R. Recker, J. Cheng, and H.W. Deng, *Whole genome distribution and ethnic differentiation of copy number variation in Caucasian and Asian populations.* PLoS One, 2009. **4**(11): p. e7958.
 44. Liu, X.G., L.J. Tan, S.F. Lei, Y.J. Liu, H. Shen, L. Wang, H. Yan, Y.F. Guo, D.H. Xiong, X.D. Chen, F. Pan, T.L. Yang, Y.P. Zhang, Y. Guo, N.L. Tang, X.Z. Zhu, H.Y. Deng, S. Levy, R.R. Recker, C.J. Papasian, and H.W. Deng, *Genome-wide association and replication studies identified TRHR as an important gene for lean body mass.* Am J Hum Genet, 2009. **84**(3): p. 418-23.
 45. Liu, Y.Z., Y.F. Guo, L. Wang, L.J. Tan, X.G. Liu, Y.F. Pei, H. Yan, D.H. Xiong, F.Y. Deng, N. Yu, Y.P. Zhang, L. Zhang, S.F. Lei, X.D. Chen, H.B. Liu, X.Z. Zhu, S. Levy, C.J. Papasian, B.M. Drees, J.J. Hamilton, R.R. Recker, and H.W. Deng, *Genome-wide association analyses identify SPOCK as a key novel gene underlying age at menarche.* PLoS Genet, 2009. **5**(3): p. e1000420.
 46. Liu, Y.Z., Y.F. Pei, Y.F. Guo, L. Wang, X.G. Liu, H. Yan, D.H. Xiong, Y.P. Zhang, S. Levy, J. Li, C.K. Haddock, C.J. Papasian, Q. Xu, J.Z. Ma, T.J. Payne, R.R. Recker, M.D. Li, and H.W. Deng, *Genome-wide association analyses suggested a novel mechanism for smoking behavior regulated by IL15.* Mol Psychiatry, 2009. **14**(7): p. 668-80.
 47. Liu, Y.Z., Y.F. Pei, J.F. Liu, F. Yang, Y. Guo, L. Zhang, X.G. Liu, H. Yan, L. Wang, Y.P. Zhang, S. Levy, R.R. Recker, and H.W. Deng, *Powerful bivariate genome-wide association analyses suggest the SOX6 gene influencing both obesity and osteoporosis phenotypes in males.* PLoS One, 2009. **4**(8): p. e6827.
 48. Xiong, D.H., X.G. Liu, Y.F. Guo, L.J. Tan, L. Wang, B.Y. Sha, Z.H. Tang, F. Pan, T.L. Yang, X.D. Chen, S.F. Lei, L.M. Yerges, X.Z. Zhu, V.W. Wheeler, A.L. Patrick, C.H. Bunker, Y. Guo, H. Yan, Y.F. Pei, Y.P. Zhang, S. Levy, C.J. Papasian, P. Xiao, Y.W. Lundberg, R.R. Recker, Y.Z. Liu, Y.J. Liu, J.M. Zmuda, and H.W. Deng, *Genome-wide association and follow-up replication studies identified ADAMTS18 and TGFBR3 as bone mass candidate genes in different ethnic groups.* Am J Hum Genet, 2009. **84**(3): p. 388-98.
 49. Zheng, W., J. Long, Y.T. Gao, C. Li, Y. Zheng, Y.B. Xiang, W. Wen, S. Levy, S.L. Deming, J.L. Haines, K. Gu, A.M. Fair, Q. Cai, W. Lu, and X.O. Shu, *Genome-wide association study identifies a new breast cancer susceptibility locus at 6q25.1.* Nat Genet, 2009. **41**(3): p. 324-8.
 50. Canter, J.A., G.K. Robbins, D. Selph, D.B. Clifford, A.R. Kallianpur, R. Shafer, S. Levy, D.G. Murdock, M.D. Ritchie, D.W. Haas, and T. Hulan, *African mitochondrial DNA subhaplogroups and peripheral neuropathy during antiretroviral therapy.* J Infect Dis, 2010. **201**(11): p. 1703-7.
 51. Greco, J.A., 3rd, A.C. Pollins, B.E. Boone, S.E. Levy, and L.B. Nannery, *A microarray analysis of temporal gene expression profiles in thermally injured human skin.* Burns, 2010. **36**(2): p. 192-204.
 52. Lange, L.A., D.C. Croteau-Chonka, A.F. Marvelle, L. Qin, K.J. Gaulton, C.W. Kuzawa, T.W. McDade, Y. Wang, Y. Li, S. Levy, J.B. Borja, E.M. Lange, L.S. Adair, and K.L. Mohlke, *Genome-wide association study of homocysteine levels in Filipinos provides evidence for CPS1 in women and a stronger MTHFR effect in young adults.* Hum Mol Genet, 2010. **19**(10): p. 2050-8.
 53. Motsinger-Reif, A.A., P.R. Antas, N.O. Oki, S. Levy, S.M. Holland, and T.R. Sterling, *Polymorphisms in IL-1beta, vitamin D receptor Fok1, and Toll-like receptor 2 are associated with extrapulmonary tuberculosis.* BMC Med Genet, 2010. **11**: p. 37.

54. Otto, E.A., T.W. Hurd, R. Airik, M. Chaki, W. Zhou, C. Stoetzel, S.B. Patil, S. Levy, A.K. Ghosh, C.A. Murga-Zamalloa, J. van Reeuwijk, S.J. Letteboer, L. Sang, R.H. Giles, Q. Liu, K.L. Coene, A. Estrada-Cuzcano, R.W. Collin, H.M. McLaughlin, S. Held, J.M. Kasanuki, G. Ramaswami, J. Conte, I. Lopez, J. Washburn, J. Macdonald, J. Hu, Y. Yamashita, E.R. Maher, L.M. Guay-Woodford, H.P. Neumann, N. Obermuller, R.K. Koenekoop, C. Bergmann, X. Bei, R.A. Lewis, N. Katsanis, V. Lopes, D.S. Williams, R.H. Lyons, C.V. Dang, D.A. Brito, M.B. Dias, X. Zhang, J.D. Cavalcoli, G. Nurnberg, P. Nurnberg, E.A. Pierce, P.K. Jackson, C. Antignac, S. Saunier, R. Roepman, H. Dollfus, H. Khanna, and F. Hildebrandt, *Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy*. Nat Genet, 2010. **42**(10): p. 840-50.
55. Smith, J.J., N.G. Deane, F. Wu, N.B. Merchant, B. Zhang, A. Jiang, P. Lu, J.C. Johnson, C. Schmidt, C.E. Bailey, S. Eschrich, C. Kis, S. Levy, M.K. Washington, M.J. Heslin, R.J. Coffey, T.J. Yeatman, Y. Shyr, and R.D. Beauchamp, *Experimentally derived metastasis gene expression profile predicts recurrence and death in patients with colon cancer*. Gastroenterology, 2010. **138**(3): p. 958-68.
56. Tan, L., R. Liu, S. Lei, R. Pan, T. Yang, H. Yan, Y. Pei, F. Yang, F. Zhang, F. Pan, Y. Zhang, H. Hu, S. Levy, and H. Deng, *A genome-wide association analysis implicates SOX6 as a candidate gene for wrist bone mass*. Sci China Life Sci, 2010. **53**(9): p. 1065-72.
57. Beane, J., J. Vick, F. Schembri, C. Anderlind, A. Gower, J. Campbell, L. Luo, X.H. Zhang, J. Xiao, Y.O. Alekseyev, S. Wang, S. Levy, P.P. Massion, M. Lenburg, and A. Spira, *Characterizing the impact of smoking and lung cancer on the airway transcriptome using RNA-Seq*. Cancer Prev Res (Phila), 2011. **4**(6): p. 803-17.
58. Cai, Q., J. Long, W. Lu, S. Qu, W. Wen, D. Kang, J.Y. Lee, K. Chen, H. Shen, C.Y. Shen, H. Sung, K. Matsuo, C.A. Haiman, U.S. Khoo, Z. Ren, M. Iwasaki, K. Gu, Y.B. Xiang, J.Y. Choi, S.K. Park, L. Zhang, Z. Hu, P.E. Wu, D.Y. Noh, K. Tajima, B.E. Henderson, K.Y. Chan, F. Su, Y. Kasuga, W. Wang, J.R. Cheng, K.Y. Yoo, J.Y. Lee, H. Zheng, Y. Liu, Y.L. Shieh, S.W. Kim, J.W. Lee, H. Iwata, L. Le Marchand, S.Y. Chan, X. Xie, S. Tsugane, M.H. Lee, S. Wang, G. Li, S. Levy, B. Huang, J. Shi, R. Delahanty, Y. Zheng, C. Li, Y.T. Gao, X.O. Shu, and W. Zheng, *Genome-wide association study identifies breast cancer risk variant at 10q21.2: results from the Asia Breast Cancer Consortium*. Hum Mol Genet, 2011. **20**(24): p. 4991-9.
59. Oki, N.O., A.A. Motsinger-Reif, P.R. Antas, S. Levy, S.M. Holland, and T.R. Sterling, *Novel human genetic variants associated with extrapulmonary tuberculosis: a pilot genome wide association study*. BMC Res Notes, 2011. **4**: p. 28.
60. Xu, B., J.L. Roos, P. Dexheimer, B. Boone, B. Plummer, S. Levy, J.A. Gogos, and M. Karayiorgou, *Exome sequencing supports a de novo mutational paradigm for schizophrenia*. Nat Genet, 2011. **43**(9): p. 864-8.
61. Arrington, C.B., S.B. Bleyl, N. Matsunami, G.D. Bonnell, B.E. Otterud, D.C. Nielsen, J. Stevens, S. Levy, M.F. Leppert, and N.E. Bowles, *Exome analysis of a family with pleiotropic congenital heart disease*. Circ Cardiovasc Genet, 2012. **5**(2): p. 175-82.
62. Chaki, M., R. Airik, A.K. Ghosh, R.H. Giles, R. Chen, G.G. Slaats, H. Wang, T.W. Hurd, W. Zhou, A. Cluckey, H.Y. Gee, G. Ramaswami, C.J. Hong, B.A. Hamilton, I. Cervenka, R.S. Ganji, V. Bryja, H.H. Arts, J. van Reeuwijk, M.M. Oud, S.J. Letteboer, R. Roepman, H. Husson, O. Ibraghimov-Beskrovnya, T. Yasunaga, G. Walz, L. Eley, J.A. Sayer, B. Schermer, M.C. Liebau, T. Benzing, S. Le Corre, I. Drummond, S. Janssen, S.J. Allen, S. Natarajan, J.F. O'Toole, M. Attanasio, S. Saunier, C. Antignac, R.K. Koenekoop, H. Ren, I. Lopez, A. Nayir, C. Stoetzel, H. Dollfus, R. Massoudi, J.G. Gleeson, S.P. Andreoli, D.G. Doherty, A. Lindstrad, C. Golzio, N. Katsanis, L. Pape, E.B. Abboud, A.A. Al-Rajhi, R.A. Lewis, H. Omran, E.Y. Lee, S. Wang, J.M. Sekiguchi, R. Saunders, C.A. Johnson, E. Garner, K. Vanselow, J.S. Andersen, J. Shlomal, G. Nurnberg, P. Nurnberg, S. Levy, A.

- Smogorzewska, E.A. Otto, and F. Hildebrandt, *Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling*. *Cell*, 2012. **150**(3): p. 533-48.
63. Chen, X.D., D.H. Xiong, T.L. Yang, Y.F. Pei, Y.F. Guo, J. Li, F. Yang, F. Pan, L.J. Tan, H. Yan, X.G. Liu, S.F. Lei, X. Li, L.L. Ning, X.Z. Zhu, S. Levy, H.R. Kranzler, L.A. Farrer, J. Gelernter, R.R. Recker, and H.W. Deng, *ANKRD7 and CYTL1 are novel risk genes for alcohol drinking behavior*. *Chin Med J (Engl)*, 2012. **125**(6): p. 1127-34.
64. Ding, D., N.M. Scott, E.E. Thompson, T. Chaiworapongsa, R. Torres, C. Billstrand, K. Murray, P.J. Dexheimer, M. Ismail, H. Kay, S. Levy, R. Romero, M.D. Lindheimer, D.L. Nicolae, and C. Ober, *Increased protein-coding mutations in the mitochondrial genome of African American women with preeclampsia*. *Reprod Sci*, 2012. **19**(12): p. 1343-51.
65. Fiskerstrand, T., N. Arshad, B.I. Haukanes, R.R. Tronstad, K.D. Pham, S. Johansson, B. Havik, S.L. Tonder, S.E. Levy, D. Brackman, H. Boman, K.H. Biswas, J. Apold, N. Hovdenak, S.S. Visweswariah, and P.M. Knappskog, *Familial diarrhea syndrome caused by an activating GUCY2C mutation*. *N Engl J Med*, 2012. **366**(17): p. 1586-95.
66. Guo, Y., Q. Cai, D.C. Samuels, F. Ye, J. Long, C.I. Li, J.F. Winther, E.J. Tawn, M. Stovall, P. Lahteenmaki, N. Malila, S. Levy, C. Shaffer, Y. Shyr, X.O. Shu, and J.D. Boice, Jr., *The use of next generation sequencing technology to study the effect of radiation therapy on mitochondrial DNA mutation*. *Mutat Res*, 2012. **744**(2): p. 154-60.
67. Johansson, S., H. Irgens, K.K. Chudasama, J. Molnes, J. Aerts, F.S. Roque, I. Jonassen, S. Levy, K. Lima, P.M. Knappskog, G.I. Bell, A. Molven, and P.R. Njolstad, *Exome sequencing and genetic testing for MODY*. *PLoS One*, 2012. **7**(5): p. e38050.
68. Love, C., Z. Sun, D. Jima, G. Li, J. Zhang, R. Miles, K.L. Richards, C.H. Dunphy, W.W. Choi, G. Srivastava, P.L. Lugar, D.A. Rizzieri, A.S. Lagoo, L. Bernal-Mizrachi, K.P. Mann, C.R. Flowers, K.N. Naresh, A.M. Evens, A. Chadburn, L.I. Gordon, M.B. Czader, J.I. Gill, E.D. Hsi, A. Greenough, A.B. Moffitt, M. McKinney, A. Banerjee, V. Grubor, S. Levy, D.B. Dunson, and S.S. Dave, *The genetic landscape of mutations in Burkitt lymphoma*. *Nat Genet*, 2012. **44**(12): p. 1321-5.
69. Neale, B.M., Y. Kou, L. Liu, A. Ma'ayan, K.E. Samocha, A. Sabo, C.F. Lin, C. Stevens, L.S. Wang, V. Makarov, P. Polak, S. Yoon, J. Maguire, E.L. Crawford, N.G. Campbell, E.T. Geller, O. Valladares, C. Schafer, H. Liu, T. Zhao, G. Cai, J. Lihm, R. Dannenfels, O. Jabado, Z. Peralta, U. Nagaswamy, D. Muzny, J.G. Reid, I. Newsham, Y. Wu, L. Lewis, Y. Han, B.F. Voight, E. Lim, E. Rossin, A. Kirby, J. Flannick, M. Fromer, K. Shakir, T. Fennell, K. Garimella, E. Banks, R. Poplin, S. Gabriel, M. DePristo, J.R. Wimbish, B.E. Boone, S.E. Levy, C. Betancur, S. Sunyaev, E. Boerwinkle, J.D. Buxbaum, E.H. Cook, Jr., B. Devlin, R.A. Gibbs, K. Roeder, G.D. Schellenberg, J.S. Sutcliffe, and M.J. Daly, *Patterns and rates of exonic de novo mutations in autism spectrum disorders*. *Nature*, 2012. **485**(7397): p. 242-5.
70. Powell, A.E., Y. Wang, Y. Li, E.J. Poulin, A.L. Means, M.K. Washington, J.N. Higginbotham, A. Juchheim, N. Prasad, S.E. Levy, Y. Guo, Y. Shyr, B.J. Aronow, K.M. Haigis, J.L. Franklin, and R.J. Coffey, *The pan-ErbB negative regulator Lrig1 is an intestinal stem cell marker that functions as a tumor suppressor*. *Cell*, 2012. **149**(1): p. 146-58.
71. Rademakers, R., M. Baker, A.M. Nicholson, N.J. Rutherford, N. Finch, A. Soto-Ortolaza, J. Lash, C. Wider, A. Wojtas, M. DeJesus-Hernandez, J. Adamson, N. Kouri, C. Sundal, E.A. Shuster, J. Aasly, J. MacKenzie, S. Roeber, H.A. Kretschmar, B.F. Boeve, D.S. Knopman, R.C. Petersen, N.J. Cairns, B. Ghetti, S. Spina, J. Garbern, A.C. Tselis, R. Uitti, P. Das, J.A. Van Gerpen, J.F. Meschia, S. Levy, D.F. Broderick, N. Graff-Radford, O.A. Ross, B.B. Miller, R.H. Swerdlow, D.W. Dickson, and Z.K. Wszolek, *Mutations in the colony stimulating factor 1 receptor (CSF1R) gene cause hereditary diffuse leukoencephalopathy with spheroids*. *Nat Genet*, 2012. **44**(2): p. 200-5.

72. Su, Y., A.E. Vilgelm, M.C. Kelley, O.E. Hawkins, Y. Liu, K.L. Boyd, S. Kantrow, R.C. Splittgerber, S.P. Short, T. Sobolik, S. Zaja-Milatovic, K.B. Dahlman, K.I. Amiri, A. Jiang, P. Lu, Y. Shyr, D.D. Stuart, S. Levy, J.A. Sosman, and A. Richmond, *RAF265 inhibits the growth of advanced human melanoma tumors*. Clin Cancer Res, 2012. **18**(8): p. 2184-98.
73. The ENCODE Project Consortium, C., *An integrated encyclopedia of DNA elements in the human genome*. Nature, 2012. **489**(7414): p. 57-74.
74. Xu, B., I. Ionita-Laza, J.L. Roos, B. Boone, S. Woodrick, Y. Sun, S. Levy, J.A. Gogos, and M. Karayiorgou, *De novo gene mutations highlight patterns of genetic and neural complexity in schizophrenia*. Nat Genet, 2012. **44**(12): p. 1365-9.
75. Zhou, W., E.A. Otto, A. Cluckey, R. Airik, T.W. Hurd, M. Chaki, K. Diaz, F.P. Lach, G.R. Bennett, H.Y. Gee, A.K. Ghosh, S. Natarajan, S. Thongthip, U. Veturi, S.J. Allen, S. Janssen, G. Ramaswami, J. Dixon, F. Burkhalter, M. Spoendlin, H. Moch, M.J. Mihatsch, J. Verine, R. Reade, H. Soliman, M. Godin, D. Kiss, G. Monga, G. Mazzucco, K. Amann, F. Artunc, R.C. Newland, T. Wiech, S. Zschiedrich, T.B. Huber, A. Friedl, G.G. Slaats, J.A. Joles, R. Goldschmeding, J. Washburn, R.H. Giles, S. Levy, A. Smogorzewska, and F. Hildebrandt, *FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair*. Nat Genet, 2012. **44**(8): p. 910-5.
76. Ashraf, S., H.Y. Gee, S. Woerner, L.X. Xie, V. Vega-Warner, S. Lovric, H. Fang, X. Song, D.C. Cattran, C. Avila-Casado, A.D. Paterson, P. Nitschke, C. Bole-Feysot, P. Cochat, J. Esteve-Rudd, B. Haberberger, S.J. Allen, W. Zhou, R. Airik, E.A. Otto, M. Barua, M.H. Al-Hamed, J.A. Kari, J. Evans, A. Bierzynska, M.A. Saleem, D. Bockenbauer, R. Kleta, S. El Desoky, D.O. Hacihamdioglu, F. Gok, J. Washburn, R.C. Wiggins, M. Choi, R.P. Lifton, S. Levy, Z. Han, L. Salviati, H. Prokisch, D.S. Williams, M. Pollak, C.F. Clarke, Y. Pei, C. Antignac, and F. Hildebrandt, *ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption*. J Clin Invest, 2013. **123**(12): p. 5179-89.
77. Chiu, I.M., E.T. Morimoto, H. Goodarzi, J.T. Liao, S. O'Keeffe, H.P. Phatnani, M. Muratet, M.C. Carroll, S. Levy, S. Tavazoie, R.M. Myers, and T. Maniatis, *A neurodegeneration-specific gene-expression signature of acutely isolated microglia from an amyotrophic lateral sclerosis mouse model*. Cell Rep, 2013. **4**(2): p. 385-401.
78. Franklin, J.L., C.R. Rankin, S. Levy, J.R. Snoddy, B. Zhang, M.K. Washington, J.M. Thomson, R.H. Whitehead, and R.J. Coffey, *Malignant transformation of colonic epithelial cells by a colon-derived long noncoding RNA*. Biochem Biophys Res Commun, 2013. **440**(1): p. 99-104.
79. Gee, H.Y., P. Saisawat, S. Ashraf, T.W. Hurd, V. Vega-Warner, H. Fang, B.B. Beck, O. Gribouval, W. Zhou, K.A. Diaz, S. Natarajan, R.C. Wiggins, S. Lovric, G. Chernin, D.S. Schoeb, B. Ovunc, Y. Frishberg, N.A. Soliman, H.M. Fathy, H. Goebel, J. Hoefele, L.T. Weber, J.W. Innis, C. Faul, Z. Han, J. Washburn, C. Antignac, S. Levy, E.A. Otto, and F. Hildebrandt, *ARHGDI1 mutations cause nephrotic syndrome via defective RHO GTPase signaling*. J Clin Invest, 2013. **123**(8): p. 3243-53.
80. Pekow, J., U. Dougherty, Y. Huang, E. Gometz, J. Nathanson, G. Cohen, S. Levy, M. Kocherginsky, N. Venu, M. Westerhoff, J. Hart, A.E. Noffsinger, S.B. Hanauer, R.D. Hurst, A. Fichera, L.J. Joseph, Q. Liu, and M. Bissonnette, *Gene signature distinguishes patients with chronic ulcerative colitis harboring remote neoplastic lesions*. Inflamm Bowel Dis, 2013. **19**(3): p. 461-70.
81. Ramirez, A.H., C.M. Shaffer, J.T. Delaney, D.P. Sexton, S.E. Levy, M.J. Rieder, D.A. Nickerson, A.L. George, Jr., and D.M. Roden, *Novel rare variants in congenital cardiac arrhythmia genes are frequent in drug-induced torsades de pointes*. Pharmacogenomics J, 2013. **13**(4): p. 325-9.
82. Walter, R.B., G.S. Laszlo, T.A. Alonzo, R.B. Gerbing, S. Levy, M.P. Fitzgibbon, C.J. Gudgeon, R.E. Ries, K.H. Harrington, S.C. Raimondi, B.A. Hirsch, A.S. Gamis, W.M. M,

- and S. Meshinchi, *Significance of expression of ITGA5 and its splice variants in acute myeloid leukemia: a report from the Children's Oncology Group*. Am J Hematol, 2013. **88**(8): p. 694-702.
83. Zariwala, M.A., H.Y. Gee, M. Kurkowiak, D.A. Al-Mutairi, M.W. Leigh, T.W. Hurd, R. Hjeij, S.D. Dell, M. Chaki, G.W. Dougherty, M. Adan, P.C. Spear, J. Esteve-Rudd, N.T. Loges, M. Rosenfeld, K.A. Diaz, H. Olbrich, W.E. Wolf, E. Sheridan, T.F. Batten, J. Halbritter, J.D. Porath, S. Kohl, S. Lovric, D.Y. Hwang, J.E. Pittman, K.A. Burns, T.W. Ferkol, S.D. Sagel, K.N. Olivier, L.C. Morgan, C. Werner, J. Raidt, P. Pennekamp, Z. Sun, W. Zhou, R. Airik, S. Natarajan, S.J. Allen, I. Amirav, D. Wieczorek, K. Landwehr, K. Nielsen, N. Schwerk, J. Sertic, G. Kohler, J. Washburn, S. Levy, S. Fan, C. Koerner-Rettberg, S. Amselem, D.S. Williams, B.J. Mitchell, I.A. Drummond, E.A. Otto, H. Omran, M.R. Knowles, and F. Hildebrandt, *ZMYND10 is mutated in primary ciliary dyskinesia and interacts with LRRC6*. Am J Hum Genet, 2013. **93**(2): p. 336-45.
 84. Zhang, J., V. Grubor, C.L. Love, A. Banerjee, K.L. Richards, P.A. Mieczkowski, C. Dunphy, W. Choi, W.Y. Au, G. Srivastava, P.L. Lugar, D.A. Rizzieri, A.S. Lagoo, L. Bernal-Mizrachi, K.P. Mann, C. Flowers, K. Naresh, A. Evens, L.I. Gordon, M. Czader, J.I. Gill, E.D. Hsi, Q. Liu, A. Fan, K. Walsh, D. Jima, L.L. Smith, A.J. Johnson, J.C. Byrd, M.A. Luftig, T. Ni, J. Zhu, A. Chadburn, S. Levy, D. Dunson, and S.S. Dave, *Genetic heterogeneity of diffuse large B-cell lymphoma*. Proc Natl Acad Sci U S A, 2013. **110**(4): p. 1398-403.
 85. Brissova, M., K. Aamodt, P. Brahmachary, N. Prasad, J.Y. Hong, C. Dai, M. Mellati, A. Shostak, G. Poffenberger, R. Aramandla, S.E. Levy, and A.C. Powers, *Islet microenvironment, modulated by vascular endothelial growth factor-A signaling, promotes beta cell regeneration*. Cell Metab, 2014. **19**(3): p. 498-511.
 86. Campbell, A.G., P. Schwientek, T. Vishnivetskaya, T. Woyke, S. Levy, C.J. Beall, A. Griffen, E. Leys, and M. Podar, *Diversity and genomic insights into the uncultured Chloroflexi from the human microbiota*. Environ Microbiol, 2014. **16**(9): p. 2635-43.
 87. Gee, H.Y., S. Ashraf, X. Wan, V. Vega-Warner, J. Esteve-Rudd, S. Lovric, H. Fang, T.W. Hurd, C.E. Sadowski, S.J. Allen, E.A. Otto, E. Korkmaz, J. Washburn, S. Levy, D.S. Williams, S.A. Bakkaloglu, A. Zolotnitskaya, F. Ozaltin, W. Zhou, and F. Hildebrandt, *Mutations in EMP2 cause childhood-onset nephrotic syndrome*. Am J Hum Genet, 2014. **94**(6): p. 884-90.
 88. Gee, H.Y., E.A. Otto, T.W. Hurd, S. Ashraf, M. Chaki, A. Cluckey, V. Vega-Warner, P. Saisawat, K.A. Diaz, H. Fang, S. Kohl, S.J. Allen, R. Airik, W. Zhou, G. Ramaswami, S. Janssen, C. Fu, J.L. Innis, S. Weber, U. Vester, E.E. Davis, N. Katsanis, H.M. Fathy, N. Jeck, G. Klaus, A. Nayir, K.A. Rahim, I. Al Attrach, I. Al Hassoun, S. Ozturk, D. Drozd, U. Helmchen, J.F. O'Toole, M. Attanasio, R.A. Lewis, G. Nurnberg, P. Nurnberg, J. Washburn, J. MacDonald, J.W. Innis, S. Levy, and F. Hildebrandt, *Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies*. Kidney Int, 2014. **85**(4): p. 880-7.
 89. Li, S., S.W. Tighe, C.M. Nicolet, D. Grove, S. Levy, W. Farmerie, A. Viale, C. Wright, P.A. Schweitzer, Y. Gao, D. Kim, J. Boland, B. Hicks, R. Kim, S. Chhangawala, N. Jafari, N. Raghavachari, J. Gandara, N. Garcia-Reyero, C. Hendrickson, D. Roberson, J. Rosenfeld, T. Smith, J.G. Underwood, M. Wang, P. Zumbo, D.A. Baldwin, G.S. Grills, and C.E. Mason, *Multi-platform assessment of transcriptome profiling using RNA-seq in the ABRF next-generation sequencing study*. Nat Biotechnol, 2014. **32**(9): p. 915-25.
 90. Ning, M.S., A.S. Kim, N. Prasad, S.E. Levy, H. Zhang, and T. Andl, *Characterization of the Merkel Cell Carcinoma miRNome*. J Skin Cancer, 2014. **2014**: p. 289548.
 91. Picard, M., J. Zhang, S. Hancock, O. Derbeneva, R. Golhar, P. Golik, S. O'Hearn, S. Levy, P. Potluri, M. Lvova, A. Davila, C.S. Lin, J.C. Perin, E.F. Rappaport, H. Hakonarson, I.A. Trounce, V. Procaccio, and D.C. Wallace, *Progressive increase in*

- mtDNA 3243A>G heteroplasmy causes abrupt transcriptional reprogramming*. Proc Natl Acad Sci U S A, 2014. **111**(38): p. E4033-42.
92. Reinert, R.B., Q. Cai, J.Y. Hong, J.L. Plank, K. Aamodt, N. Prasad, R. Aramandla, C. Dai, S.E. Levy, A. Pozzi, P.A. Labosky, C.V. Wright, M. Brissova, and A.C. Powers, *Vascular endothelial growth factor coordinates islet innervation via vascular scaffolding*. Development, 2014. **141**(7): p. 1480-91.
 93. Royse, K.E., D. Zhi, M.G. Conner, B. Clodfelder-Miller, V. Srinivasasainagendra, L.K. Vaughan, C.F. Skibola, D.K. Crossman, S. Levy, and S. Shrestha, *Differential Gene Expression Landscape of Co-Existing Cervical Pre-Cancer Lesions Using RNA-seq*. Front Oncol, 2014. **4**: p. 339.
 94. Vrieze, S.I., S.M. Malone, U. Vaidyanathan, A. Kwong, H.M. Kang, X. Zhan, M. Flickinger, D. Irons, G. Jun, A.E. Locke, G. Pistis, E. Porcu, S. Levy, R.M. Myers, W. Oetting, M. McGue, G. Abecasis, and W.G. Iacono, *In search of rare variants: preliminary results from whole genome sequencing of 1,325 individuals with psychophysiological endophenotypes*. Psychophysiology, 2014. **51**(12): p. 1309-20.
 95. Zhang, J., D. Jima, A.B. Moffitt, Q. Liu, M. Czader, E.D. Hsi, Y. Fedoriw, C.H. Dunphy, K.L. Richards, J.I. Gill, Z. Sun, C. Love, P. Scotland, E. Lock, S. Levy, D.S. Hsu, D. Dunson, and S.S. Dave, *The genomic landscape of mantle cell lymphoma is related to the epigenetically determined chromatin state of normal B cells*. Blood, 2014. **123**(19): p. 2988-96.
 96. Afshinnekoo, E., C. Meydan, S. Chowdhury, D. Jaroudi, C. Boyer, N. Bernstein, J.M. Maritz, D. Reeves, J. Gandara, S. Chhangawala, S. Ahsanuddin, A. Simmons, T. Nessel, B. Sundares, E. Pereira, E. Jorgensen, S.O. Kolokotronis, N. Kirchberger, I. Garcia, D. Gandara, S. Dhanraj, T. Nawrin, Y. Saletore, N. Alexander, P. Vijay, E.M. Henaff, P. Zumbo, M. Walsh, G.D. O'Mullan, S. Tighe, J.T. Dudley, A. Dunaif, S. Ennis, E. O'Halloran, T.R. Magalhaes, B. Boone, A.L. Jones, T.R. Muth, K.S. Paolantonio, E. Alter, E.E. Schadt, J. Garbarino, R.J. Prill, J.M. Carlton, S. Levy, and C.E. Mason, *Geospatial Resolution of Human and Bacterial Diversity with City-Scale Metagenomics*. Cell Syst, 2015. **1**(1): p. 72-87.
 97. Cha, D.J., J.L. Franklin, Y. Dou, Q. Liu, J.N. Higginbotham, M. Demory Beckler, A.M. Weaver, K. Vickers, N. Prasad, S. Levy, B. Zhang, R.J. Coffey, and J.G. Patton, *KRAS-dependent sorting of miRNA to exosomes*. Elife, 2015. **4**: p. e07197.
 98. Cirulli, E.T., B.N. Lasseigne, S. Petrovski, P.C. Sapp, P.A. Dion, C.S. Leblond, J. Couthouis, Y.F. Lu, Q. Wang, B.J. Krueger, Z. Ren, J. Keebler, Y. Han, S.E. Levy, B.E. Boone, J.R. Wimbish, L.L. Waite, A.L. Jones, J.P. Carulli, A.G. Day-Williams, J.F. Staropoli, W.W. Xin, A. Chesi, A.R. Raphael, D. McKenna-Yasek, J. Cady, J.M. Vianney de Jong, K.P. Kenna, B.N. Smith, S. Topp, J. Miller, A. Gkazi, A. Al-Chalabi, L.H. van den Berg, J. Veldink, V. Silani, N. Ticozzi, C.E. Shaw, R.H. Baloh, S. Appel, E. Simpson, C. Lagier-Tourenne, S.M. Pulst, S. Gibson, J.Q. Trojanowski, L. Elman, L. McCluskey, M. Grossman, N.A. Shneider, W.K. Chung, J.M. Ravits, J.D. Glass, K.B. Sims, V.M. Van Deerlin, T. Maniatis, S.D. Hayes, A. Ordureau, S. Swarup, J. Landers, F. Baas, A.S. Allen, R.S. Bedlack, J.W. Harper, A.D. Gitler, G.A. Rouleau, R. Brown, M.B. Harms, G.M. Cooper, T. Harris, R.M. Myers, and D.B. Goldstein, *Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways*. Science, 2015. **347**(6229): p. 1436-41.
 99. Ebarasi, L., S. Ashraf, A. Bierzynska, H.Y. Gee, H.J. McCarthy, S. Lovric, C.E. Sadowski, W. Pabst, V. Vega-Warner, H. Fang, A. Koziell, M.A. Simpson, I. Dursun, E. Serdaroglu, S. Levy, M.A. Saleem, F. Hildebrandt, and A. Majumdar, *Defects of CRB2 cause steroid-resistant nephrotic syndrome*. Am J Hum Genet, 2015. **96**(1): p. 153-61.
 100. Gee, H.Y., F. Zhang, S. Ashraf, S. Kohl, C.E. Sadowski, V. Vega-Warner, W. Zhou, S. Lovric, H. Fang, M. Nettleton, J.Y. Zhu, J. Hoefele, L.T. Weber, L. Podracka, A. Boor, H.

- Fehrenbach, J.W. Innis, J. Washburn, S. Levy, R.P. Lifton, E.A. Otto, Z. Han, and F. Hildebrandt, *KANK deficiency leads to podocyte dysfunction and nephrotic syndrome*. J Clin Invest, 2015. **125**(6): p. 2375-84.
101. Hoek, K.L., P. Samir, L.M. Howard, X. Niu, N. Prasad, A. Galassie, Q. Liu, T.M. Allos, K.A. Floyd, Y. Guo, Y. Shyr, S.E. Levy, S. Joyce, K.M. Edwards, and A.J. Link, *A cell-based systems biology assessment of human blood to monitor immune responses after influenza vaccination*. PLoS One, 2015. **10**(2): p. e0118528.
102. Hsu, C.H., C. Nguyen, C. Yan, R.E. Ries, Q.R. Chen, Y. Hu, F. Ostronoff, D.L. Stirewalt, G. Komatsoulis, S. Levy, D. Meerzaman, and S. Meshinchi, *Transcriptome Profiling of Pediatric Core Binding Factor AML*. PLoS One, 2015. **10**(9): p. e0138782.
103. Rahman, K.M., M.E. Camp, N. Prasad, A.K. McNeel, S.E. Levy, F.F. Bartol, and C.A. Bagnell, *Age and Nursing Affect the Neonatal Porcine Uterine Transcriptome*. Biol Reprod, 2015.
104. Strong, K.A., T. May, and S.A. Levy, *In sickness and in health: context matters when considering potential benefits and risks of genome-wide sequencing*. Genet Med, 2015. **17**(8): p. 681-2.
105. Zhi, D., A. Shendure, R. Scherzer, M.R. Irvin, R.T. Perry, S. Levy, D.K. Arnett, C. Grunfeld, and S. Shrestha, *Deep sequencing of RYR3 gene identifies rare and common variants associated with increased carotid intima-media thickness (cIMT) in HIV-infected individuals*. J Hum Genet, 2015. **60**(2): p. 63-7.
106. Alam, S.G., Q. Zhang, N. Prasad, Y. Li, S. Chamala, R. Kuchibhotla, B. Kc, V. Aggarwal, S. Shrestha, A.L. Jones, S.E. Levy, K.J. Roux, J.A. Nickerson, and T.P. Lele, *The mammalian LINC complex regulates genome transcriptional responses to substrate rigidity*. Sci Rep, 2016. **6**: p. 38063.
107. Das, S., L. Forer, S. Schonherr, C. Sidore, A.E. Locke, A. Kwong, S.I. Vrieze, E.Y. Chew, S. Levy, M. McGue, D. Schlessinger, D. Stambolian, P.R. Loh, W.G. Iacono, A. Swaroop, L.J. Scott, F. Cucca, F. Kronenberg, M. Boehnke, G.R. Abecasis, and C. Fuchsberger, *Next-generation genotype imputation service and methods*. Nat Genet, 2016. **48**(10): p. 1284-1287.
108. Dou, Y., D.J. Cha, J.L. Franklin, J.N. Higginbotham, D.K. Jeppesen, A.M. Weaver, N. Prasad, S. Levy, R.J. Coffey, J.G. Patton, and B. Zhang, *Circular RNAs are down-regulated in KRAS mutant colon cancer cells and can be transferred to exosomes*. Sci Rep, 2016. **6**: p. 37982.
109. Goes, F.S., M. Pirooznia, J.S. Parla, M. Kramer, E. Ghiban, S. Mavruk, Y.C. Chen, E.T. Monson, V.L. Willour, R. Karchin, M. Flickinger, A.E. Locke, S.E. Levy, L.J. Scott, M. Boehnke, E. Stahl, J.L. Moran, C.M. Hultman, M. Landen, S.M. Purcell, P. Sklar, P.P. Zandi, W.R. McCombie, and J.B. Potash, *Exome Sequencing of Familial Bipolar Disorder*. JAMA Psychiatry, 2016. **73**(6): p. 590-7.
110. Graham, H.T., D.M. Rotroff, S.W. Marvel, J.B. Buse, T.M. Havener, A.G. Wilson, M.J. Wagner, A.A. Motsinger-Reif, and A.A. Investigators, *Incorporating Concomitant Medications into Genome-Wide Analyses for the Study of Complex Disease and Drug Response*. Frontiers in genetics, 2016. **7**.
111. Grimes, J.A., N. Prasad, S. Levy, R. Cattley, S. Lindley, H.W. Boothe, R.A. Henderson, and B.F. Smith, *A comparison of microRNA expression profiles from splenic hemangiosarcoma, splenic nodular hyperplasia, and normal spleens of dogs*. BMC Vet Res, 2016. **12**(1): p. 272.
112. Levy, S.E. and R.M. Myers, *Advancements in next-generation sequencing*. Annual review of genomics and human genetics, 2016. **17**: p. 95-115.
113. McCarthy, S., S. Das, W. Kretzschmar, O. Delaneau, A.R. Wood, A. Teumer, H.M. Kang, C. Fuchsberger, P. Danecek, K. Sharp, Y. Luo, C. Sidore, A. Kwong, N. Timpson, S. Koskinen, S. Vrieze, L.J. Scott, H. Zhang, A. Mahajan, J. Veldink, U. Peters, C. Pato,

- C.M. van Duijn, C.E. Gillies, I. Gandin, M. Mezzavilla, A. Gilly, M. Cocca, M. Traglia, A. Angius, J.C. Barrett, D. Boomsma, K. Branham, G. Breen, C.M. Brummett, F. Busonero, H. Campbell, A. Chan, S. Chen, E. Chew, F.S. Collins, L.J. Corbin, G.D. Smith, G. Dedoussis, M. Dorr, A.E. Farmaki, L. Ferrucci, L. Forer, R.M. Fraser, S. Gabriel, S. Levy, L. Groop, T. Harrison, A. Hattersley, O.L. Holmen, K. Hveem, M. Kretzler, J.C. Lee, M. McGue, T. Meitinger, D. Melzer, J.L. Min, K.L. Mohlke, J.B. Vincent, M. Nauck, D. Nickerson, A. Palotie, M. Pato, N. Pirastu, M. McInnis, J.B. Richards, C. Sala, V. Salomaa, D. Schlessinger, S. Schoenherr, P.E. Slagboom, K. Small, T. Spector, D. Stambolian, M. Tuke, J. Tuomilehto, L.H. Van den Berg, W. Van Rheenen, U. Volker, C. Wijmenga, D. Toniolo, E. Zeggini, P. Gasparini, M.G. Sampson, J.F. Wilson, T. Frayling, P.I. de Bakker, M.A. Swertz, S. McCarroll, C. Kooperberg, A. Dekker, D. Altshuler, C. Willer, W. Iacono, S. Ripatti, N. Soranzo, K. Walter, A. Swaroop, F. Cucca, C.A. Anderson, R.M. Myers, M. Boehnke, M.I. McCarthy, R. Durbin and C. Haplotype Reference, *A reference panel of 64,976 haplotypes for genotype imputation*. Nat Genet, 2016. **48**(10): p. 1279-83.
114. McGrath, K., L. McGrath, A. Gray, O. Osuolale, N. Segata, S. Fillo, G. Iraola, Y. Zhou, Y. Chang, and Y. Li, *The Metagenomics and Metadesign of the Subways and Urban Biomes (MetaSUB) International Consortium Inaugural Meeting Report (vol 4, 24, 2016)*. MICROBIOME, 2016. **4**.
 115. Rahman, K.M., M.E. Camp, N. Prasad, A.K. McNeel, S.E. Levy, F.F. Bartol, and C.A. Bagnell, *Age and Nursing Affect the Neonatal Porcine Uterine Transcriptome*. Biol Reprod, 2016. **94**(2): p. 46.
 116. Vilgelm, A.E., C.A. Johnson, N. Prasad, J. Yang, S.C. Chen, G.D. Ayers, J.S. Pawlikowski, D. Raman, J.A. Sosman, M. Kelley, J.A. Ecsedy, Y. Shyr, S.E. Levy, and A. Richmond, *Connecting the Dots: Therapy-Induced Senescence and a Tumor-Suppressive Immune Microenvironment*. J Natl Cancer Inst, 2016. **108**(6): p. djv406.
 117. Zea, L., N. Prasad, S.E. Levy, L. Stodieck, A. Jones, S. Shrestha, and D. Klaus, *A Molecular Genetic Basis Explaining Altered Bacterial Behavior in Space*. PLoS One, 2016. **11**(11): p. e0164359.
 118. Carlson, J., L.J. Scott, A.E. Locke, M. Flickinger, S. Levy, R.M. Myers, M. Boehnke, H.M. Kang, J.Z. Li, and S. Zöllner, *Extremely rare variants reveal patterns of germline mutation rate heterogeneity in humans*. bioRxiv, 2017: p. 108290.
 119. Chao, H.T., M. Davids, E. Burke, J.G. Pappas, J.A. Rosenfeld, A.J. McCarty, T. Davis, L. Wolfe, C. Toro, C. Tifft, F. Xia, N. Stong, T.K. Johnson, C.G. Warr, N. Undiagnosed Diseases, S. Yamamoto, D.R. Adams, T.C. Markello, W.A. Gahl, H.J. Bellen, M.F. Wangler, and M.C. Malicdan, *A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3*. Am J Hum Genet, 2017. **100**(1): p. 128-137.
 120. Dai, C., Y. Hang, A. Shostak, G. Poffenberger, N. Hart, N. Prasad, N. Phillips, S.E. Levy, D.L. Greiner, L.D. Shultz, R. Bottino, S.K. Kim, and A.C. Powers, *Age-dependent human beta cell proliferation induced by glucagon-like peptide 1 and calcineurin signaling*. J Clin Invest, 2017. **127**(10): p. 3835-3844.
 121. Dean, E.D., M. Li, N. Prasad, S.N. Wisniewski, A. Von Deylen, J. Spaeth, L. Maddison, A. Botros, L.R. Sedgeman, N. Bozadjieva, O. Ilkayeva, A. Coldren, G. Poffenberger, A. Shostak, M.C. Semich, K.I. Aamodt, N. Phillips, H. Yan, E. Bernal-Mizrachi, J.D. Corbin, K.C. Vickers, S.E. Levy, C. Dai, C. Newgard, W. Gu, R. Stein, W. Chen, and A.C. Powers, *Interrupted Glucagon Signaling Reveals Hepatic alpha Cell Axis and Role for L-Glutamine in alpha Cell Proliferation*. Cell Metab, 2017. **25**(6): p. 1362-1373 e5.
 122. Gosline, S.J., H. Weinberg, P. Knight, T. Yu, X. Guo, N. Prasad, A. Jones, S. Shrestha, B. Boone, S.E. Levy, S. La Rosa, J. Guinney, and A. Bakker, *A high-throughput molecular data resource for cutaneous neurofibromas*. Sci Data, 2017. **4**: p. 170045.

123. Howard, L.M., K.L. Hoek, J.B. Goll, P. Samir, A. Galassie, T.M. Allos, X. Niu, L.E. Gordy, C.B. Creech, N. Prasad, T.L. Jensen, H. Hill, S.E. Levy, S. Joyce, A.J. Link, and K.M. Edwards, *Cell-Based Systems Biology Analysis of Human AS03-Adjuvanted H5N1 Avian Influenza Vaccine Responses: A Phase I Randomized Controlled Trial*. PLoS One, 2017. **12**(1): p. e0167488.
124. Johansson, B.B., H.U. Irgens, J. Molnes, P. Sztromwasser, I. Aukrust, P.B. Juliusson, O. Sovik, S. Levy, T. Skrivarhaug, G. Joner, A. Molven, S. Johansson, and P.R. Njolstad, *Targeted next-generation sequencing reveals MODY in up to 6.5% of antibody-negative diabetes cases listed in the Norwegian Childhood Diabetes Registry*. Diabetologia, 2017. **60**(4): p. 625-635.
125. Keele, G.R., J.W. Prokop, H. He, K. Holl, J. Littrell, A. Deal, S. Francic, L. Cui, D.M. Gatti, K.W. Broman, M. Tschannen, S.W. Tsaih, M. Zagloul, Y. Kim, B. Baur, J. Fox, M. Robinson, S. Levy, M.J. Flister, R. Mott, W. Valdar, and L.C. Solberg Woods, *Genetic Fine-Mapping and Identification of Candidate Genes and Variants for Adiposity Traits in Outbred Rats*. Obesity (Silver Spring), 2017.
126. Luo, W., D.L. Galvan, L.E. Woodard, D. Dorset, S. Levy, and M.H. Wilson, *Comparative analysis of chimeric ZFP-, TALE- and Cas9-piggyBac transposases for integration into a single locus in human cells*. Nucleic Acids Res, 2017.
127. Mason, C., A. McIntyre, R. Ounit, E. Afshinnekoo, R. Prill, E. Henaff, N. Alexander, S. Minot, D. Danko, and J. Foox, *Comprehensive Benchmarking and Ensemble Approaches for Metagenomic Classifiers*. bioRxiv, 2017: p. 156919.
128. Mason, C.E., E. Afshinnekoo, S. Tighe, S. Wu, and S. Levy, *International Standards for Genomes, Transcriptomes, and Metagenomes*. J Biomol Tech, 2017. **28**(1): p. 8-18.
129. McDaniel, J.M., K.E. Varley, J. Gertz, D.S. Savic, B.S. Roberts, S.K. Bailey, L.A. Shevde, R.C. Ramaker, B.N. Lasseigne, M.K. Kirby, K.M. Newberry, E.C. Partridge, A.L. Jones, B. Boone, S.E. Levy, P.G. Oliver, K.C. Sexton, W.E. Grizzle, A. Forero, D.J. Buchsbaum, S.J. Cooper, and R.M. Myers, *Genomic regulation of invasion by STAT3 in triple negative breast cancer*. Oncotarget, 2017. **8**(5): p. 8226-8238.
130. McIntyre, A.B.R., R. Ounit, E. Afshinnekoo, R.J. Prill, E. Henaff, N. Alexander, S.S. Minot, D. Danko, J. Foox, S. Ahsanuddin, S. Tighe, N.A. Hasan, P. Subramanian, K. Moffat, S. Levy, S. Lonardi, N. Greenfield, R.R. Colwell, G.L. Rosen, and C.E. Mason, *Comprehensive benchmarking and ensemble approaches for metagenomic classifiers*. Genome Biol, 2017. **18**(1): p. 182.
131. McKinney, M., A.B. Moffitt, P. Gaulard, M. Travert, L. De Leval, A. Nicolae, M. Raffeld, E.S. Jaffe, S. Pittaluga, L. Xi, T. Heavican, J. Iqbal, K. Belhadj, M.H. Delfau-Larue, V. Fataccioli, M.B. Czader, I.S. Lossos, J.R. Chapman-Fredricks, K.L. Richards, Y. Fedoriw, S.L. Ondrejka, E.D. Hsi, L. Low, D. Weisenburger, W.C. Chan, N. Mehta-Shah, S. Horwitz, L. Bernal-Mizrachi, C.R. Flowers, A.W. Beaven, M. Parihar, L. Baseggio, M. Parrens, A. Moreau, P. Sujobert, M. Pilichowska, A.M. Evens, A. Chadburn, R.K. Au-Yeung, G. Srivastava, W.W. Choi, J.R. Goodlad, I. Aurer, S. Basic-Kinda, R.D. Gascoyne, N.S. Davis, G. Li, J. Zhang, D. Rajagopalan, A. Reddy, C. Love, S. Levy, Y. Zhuang, J. Datta, D.B. Dunson, and S.S. Dave, *The Genetic Basis of Hepatosplenic T-cell Lymphoma*. Cancer Discov, 2017. **7**(4): p. 369-379.
132. Moffitt, A.B., S.L. Ondrejka, M. McKinney, R.E. Rempel, J.R. Goodlad, C.H. Teh, S. Leppa, S. Mannisto, P.E. Kovanen, E. Tse, R.K.H. Au-Yeung, Y.L. Kwong, G. Srivastava, J. Iqbal, J. Yu, K. Naresh, D. Villa, R.D. Gascoyne, J. Said, M.B. Czader, A. Chadburn, K.L. Richards, D. Rajagopalan, N.S. Davis, E.C. Smith, B.C. Palus, T.J. Tzeng, J.A. Healy, P.L. Lugar, J. Datta, C. Love, S. Levy, D.B. Dunson, Y. Zhuang, E.D. Hsi, and S.S. Dave, *Enteropathy-associated T cell lymphoma subtypes are characterized by loss of function of SETD2*. J Exp Med, 2017. **214**(5): p. 1371-1386.

133. O'Hara, N.B., H.J. Reed, E. Afshinnnekoo, D. Harvin, N. Caplan, G. Rosen, B. Frye, S. Woloszynek, R. Ounit, S. Levy, E. Butler, and C.E. Mason, *Metagenomic characterization of ambulances across the USA*. Microbiome, 2017. **5**(1): p. 125.
134. Petersen, C.P., A.R. Meyer, C. De Salvo, E. Choi, C. Schlegel, A. Petersen, A.C. Engevik, N. Prasad, S.E. Levy, R.S. Peebles, T.T. Pizarro, and J.R. Goldenring, *A signalling cascade of IL-33 to IL-13 regulates metaplasia in the mouse stomach*. Gut, 2017.
135. Reddy, A., J. Zhang, N.S. Davis, A.B. Moffitt, C.L. Love, A. Waldrop, S. Leppa, A. Pasanen, L. Meriranta, M.L. Karjalainen-Lindsberg, P. Norgaard, M. Pedersen, A.O. Gang, E. Hogdall, T.B. Heavican, W. Lone, J. Iqbal, Q. Qin, G. Li, S.Y. Kim, J. Healy, K.L. Richards, Y. Fedoriw, L. Bernal-Mizrachi, J.L. Koff, A.D. Staton, C.R. Flowers, O. Paltiel, N. Goldschmidt, M. Calaminici, A. Clear, J. Gribben, E. Nguyen, M.B. Czader, S.L. Ondrejka, A. Collie, E.D. Hsi, E. Tse, R.K.H. Au-Yeung, Y.L. Kwong, G. Srivastava, W.W.L. Choi, A.M. Evens, M. Pilichowska, M. Sengar, N. Reddy, S. Li, A. Chadburn, L.I. Gordon, E.S. Jaffe, S. Levy, R. Rempel, T. Tzeng, L.E. Happ, T. Dave, D. Rajagopalan, J. Datta, D.B. Dunson, and S.S. Dave, *Genetic and Functional Drivers of Diffuse Large B Cell Lymphoma*. Cell, 2017. **171**(2): p. 481-494 e15.
136. Steffen, M.M., T.W. Davis, R.M.L. McKay, G.S. Bullerjahn, L.E. Krausfeldt, J.M.A. Stough, M.L. Neitzey, N.E. Gilbert, G.L. Boyer, T.H. Johengen, D.C. Gossiaux, A.M. Burtner, D. Palladino, M.D. Rowe, G.J. Dick, K.A. Meyer, S. Levy, B.E. Boone, R.P. Stumpf, T.T. Wynne, P.V. Zimba, D. Gutierrez, and S.W. Wilhelm, *Ecophysiological Examination of the Lake Erie Microcystis Bloom in 2014: Linkages between Biology and the Water Supply Shutdown of Toledo, OH*. Environ Sci Technol, 2017. **51**(12): p. 6745-6755.
137. Tighe, S., E. Afshinnnekoo, T.M. Rock, K. McGrath, N. Alexander, A. McIntyre, S. Ahsanuddin, D. Bezdan, S.J. Green, S. Joye, S. Stewart Johnson, D.A. Baldwin, N. Bivens, N. Ajami, J.R. Carmical, I.C. Herriott, R. Colwell, M. Donia, J. Foox, N. Greenfield, T. Hunter, J. Hoffman, J. Hyman, E. Jorgensen, D. Krawczyk, J. Lee, S. Levy, N. Garcia-Reyero, M. Settles, K. Thomas, F. Gomez, L. Schriml, N. Kyrpides, E. Zaikova, J. Penterman, and C.E. Mason, *Genomic Methods and Microbiological Technologies for Profiling Novel and Extreme Environments for the Extreme Microbiome Project (XMP)*. J Biomol Tech, 2017. **28**(1): p. 31-39.
138. Aunins, T.R., K.E. Erickson, N. Prasad, S.E. Levy, A. Jones, S. Shrestha, R. Mastracchio, L. Stodieck, D. Klaus, L. Zea, and A. Chatterjee, *Spaceflight Modifies Escherichia coli Gene Expression in Response to Antibiotic Exposure and Reveals Role of Oxidative Stress Response*. Front Microbiol, 2018. **9**: p. 310.
139. Bipolar, D., d.r.v.e. Schizophrenia Working Group of the Psychiatric Genomics Consortium. Electronic address, D. Bipolar, and C. Schizophrenia Working Group of the Psychiatric Genomics, *Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes*. Cell, 2018. **173**(7): p. 1705-1715 e16.
140. Brissova, M., R. Haliyur, D. Saunders, S. Shrestha, C. Dai, D.M. Blodgett, R. Bottino, M. Campbell-Thompson, R. Aramandla, G. Poffenberger, J. Lindner, F.C. Pan, M.G. von Herrath, D.L. Greiner, L.D. Shultz, M. Sanyoura, L.H. Philipson, M. Atkinson, D.M. Harlan, S.E. Levy, N. Prasad, R. Stein, and A.C. Powers, *alpha Cell Function and Gene Expression Are Compromised in Type 1 Diabetes*. Cell Rep, 2018. **22**(10): p. 2667-2676.
141. Da Mesquita, S., A. Louveau, A. Vaccari, I. Smirnov, R.C. Cornelison, K.M. Kingsmore, C. Contarino, S. Onengut-Gumuscu, E. Farber, D. Raper, K.E. Viar, R.D. Powell, W. Baker, N. Dabhi, R. Bai, R. Cao, S. Hu, S.S. Rich, J.M. Munson, M.B. Lopes, C.C. Overall, S.T. Acton, and J. Kipnis, *Functional aspects of meningeal lymphatics in ageing and Alzheimer's disease*. Nature, 2018. **560**(7717): p. 185-191.

142. Dickinson, S.E., B.A. Griffin, M.F. Elmore, L. Kriese-Anderson, J.B. Elmore, P.W. Dyce, S.P. Rodning, and F.H. Biase, *Transcriptome profiles in peripheral white blood cells at the time of artificial insemination discriminate beef heifers with different fertility potential*. BMC Genomics, 2018. **19**(1): p. 129.
143. Fish, E.J., K.J. Irizarry, P. Delnncientes, C.J. Ellis, N. Prasad, A.G. Moss, and R. Curt Bird, *Malignant canine mammary epithelial cells shed exosomes containing differentially expressed microRNA that regulate oncogenic networks*. BMC Cancer, 2018. **18**(1): p. 832.
144. Hiatt, S.M., M.D. Amaral, K.M. Bowling, C.R. Finnilla, M.L. Thompson, D.E. Gray, J.M.J. Lawlor, J.N. Cochran, E.M. Bebin, K.B. Brothers, K.M. East, W.V. Kelley, N.E. Lamb, S.E. Levy, E.J. Lose, M.B. Neu, C.A. Rich, S. Simmons, R.M. Myers, G.S. Barsh, and G.M. Cooper, *Systematic reanalysis of genomic data improves quality of variant interpretation*. Clin Genet, 2018. **94**(1): p. 174-178.
145. Hinger, S.A., D.J. Cha, J.L. Franklin, J.N. Higginbotham, Y. Dou, J. Ping, L. Shu, N. Prasad, S. Levy, B. Zhang, Q. Liu, A.M. Weaver, R.J. Coffey, and J.G. Patton, *Diverse Long RNAs Are Differentially Sorted into Extracellular Vesicles Secreted by Colorectal Cancer Cells*. Cell Rep, 2018. **25**(3): p. 715-725 e4.
146. Howard, L.M., J.B. Goll, T.L. Jensen, K.L. Hoek, N. Prasad, C.E. Gelber, S.E. Levy, S. Joyce, A.J. Link, C.B. Creech, and K.M. Edwards, *AS03-Adjuvanted H5N1 Avian Influenza Vaccine Modulates Early Innate Immune Signatures In Human Peripheral Blood Mononuclear Cells*. J Infect Dis, 2018.
147. Keele, G.R., J.W. Prokop, H. He, K. Holl, J. Littrell, A. Deal, S. Francic, L. Cui, D.M. Gatti, K.W. Broman, M. Tschannen, S.W. Tsaih, M. Zagloul, Y. Kim, B. Baur, J. Fox, M. Robinson, S. Levy, M.J. Flister, R. Mott, W. Valdar, and L.C. Solberg Woods, *Genetic Fine-Mapping and Identification of Candidate Genes and Variants for Adiposity Traits in Outbred Rats*. Obesity (Silver Spring), 2018. **26**(1): p. 213-222.
148. Levy, S.E. and B.E. Boone, *Next-Generation Sequencing Strategies*. Cold Spring Harb Perspect Med, 2018.
149. Liu, N., K. Schoch, X. Luo, L.D.M. Pena, V.H. Bhavana, M.K. Kukolich, S. Stringer, Z. Powis, K. Radtke, C. Mroske, K.L. Deak, M.T. McDonald, A. McConkie-Rosell, M.L. Markert, P.G. Kranz, N. Stong, A.C. Need, D. Bick, M.D. Amaral, E.A. Worthey, S. Levy, N. Undiagnosed Diseases, M.F. Wangler, H.J. Bellen, V. Shashi, and S. Yamamoto, *Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder*. Hum Mol Genet, 2018.
150. Louveau, A., J. Herz, M.N. Alme, A.F. Salvador, M.Q. Dong, K.E. Viar, S.G. Herod, J. Knopp, J.C. Setliff, A.L. Lupi, S. Da Mesquita, E.L. Frost, A. Gaultier, T.H. Harris, R. Cao, S. Hu, J.R. Lukens, I. Smirnov, C.C. Overall, G. Oliver, and J. Kipnis, *CNS lymphatic drainage and neuroinflammation are regulated by meningeal lymphatic vasculature*. Nat Neurosci, 2018.
151. Lyu, X., M.J. Rowley, and V.G. Corces, *Architectural Proteins and Pluripotency Factors Cooperate to Orchestrate the Transcriptional Response of hESCs to Temperature Stress*. Mol Cell, 2018. **71**(6): p. 940-955 e7.
152. Nicolas, A., K.P. Kenna, A.E. Renton, N. Ticozzi, F. Faghri, R. Chia, J.A. Dominov, B.J. Kenna, M.A. Nalls, P. Keagle, A.M. Rivera, W. van Rheenen, N.A. Murphy, J. van Vugt, J.T. Geiger, R.A. Van der Spek, H.A. Pliner, Shankaracharya, B.N. Smith, G. Marangi, S.D. Topp, Y. Abramzon, A.S. Gkazi, J.D. Eicher, A. Kenna, I. Consortium, G. Mora, A. Calvo, L. Mazzini, N. Riva, J. Mandrioli, C. Caponnetto, S. Battistini, P. Volanti, V. La Bella, F.L. Conforti, G. Borghero, S. Messina, I.L. Simone, F. Trojsi, F. Salvi, F.O. Logullo, S. D'Alfonso, L. Corrado, M. Capasso, L. Ferrucci, A.L.S.C.C. Genomic Translation for, C.A.M. Moreno, S. Kamalakaran, D.B. Goldstein, A.L.S.S. Consortium, A.D. Gitler, T. Harris, R.M. Myers, N.A. Consortium, H. Phatnani, R.L. Musunuri, U.S.

- Evani, A. Abhyankar, M.C. Zody, A.L.S.F. Answer, J. Kaye, S. Finkbeiner, S.K. Wyman, A. LeNail, L. Lima, E. Fraenkel, C.N. Svendsen, L.M. Thompson, J.E. Van Eyk, J.D. Berry, T.M. Miller, S.J. Kolb, M. Cudkowicz, E. Baxi, A.L.S. Clinical Research in, C. Related Disorders for Therapeutic Development, M. Benatar, J.P. Taylor, E. Rampersaud, G. Wu, J. Wu, S. Consortium, G. Lauria, F. Verde, I. Fogh, C. Tiloca, G.P. Comi, G. Soraru, C. Cereda, A.L.S.C. French, P. Corcia, H. Laaksovirta, L. Myllykangas, L. Jansson, M. Valori, J. Ealing, H. Hamdalla, S. Rollinson, S. Pickering-Brown, R.W. Orrell, K.C. Sidle, A. Malaspina, J. Hardy, A.B. Singleton, J.O. Johnson, S. Arepalli, P.C. Sapp, D. McKenna-Yasek, M. Polak, S. Asress, S. Al-Sarraj, A. King, C. Troakes, C. Vance, J. de Belleruche, F. Baas, A. Ten Asbroek, J.L. Munoz-Blanco, D.G. Hernandez, J. Ding, J.R. Gibbs, S.W. Scholz, M.K. Floeter, R.H. Campbell, F. Landi, R. Bowser, S.M. Pulst, J.M. Ravits, D.J.L. MacGowan, J. Kirby, E.P. Pioro, R. Pamphlett, J. Broach, G. Gerhard, T.L. Dunckley, C.B. Brady, N.W. Kowall, J.C. Troncoso, I. Le Ber, K. Mouzat, S. Lumbroso, T.D. Heiman-Patterson, F. Kamel, L. Van Den Bosch, R.H. Baloh, T.M. Strom, T. Meitinger, A. Shatunov, K.R. Van Eijk, M. de Carvalho, M. Kooyman, B. Middelkoop, M. Moisse, R.L. McLaughlin, M.A. Van Es, M. Weber, K.B. Boylan, M. Van Blitterswijk, R. Rademakers, K.E. Morrison, A.N. Basak, J.S. Mora, V.E. Drory, P.J. Shaw, M.R. Turner, K. Talbot, O. Hardiman, K.L. Williams, J.A. Fifita, G.A. Nicholson, I.P. Blair, G.A. Rouleau, J. Esteban-Perez, A. Garcia-Redondo, A. Al-Chalabi, E.A.L.S.S.C. Project Min, E. Rogaeva, L. Zinman, L.W. Ostrow, N.J. Maragakis, J.D. Rothstein, Z. Simmons, J. Cooper-Knock, A. Brice, S.A. Goutman, E.L. Feldman, S.B. Gibson, F. Taroni, A. Ratti, C. Gellera, P. Van Damme, W. Robberecht, P. Fratta, M. Sabatelli, C. Lunetta, A.C. Ludolph, P.M. Andersen, J.H. Weishaupt, W. Camu, J.Q. Trojanowski, V.M. Van Deerlin, R.H. Brown, Jr., L.H. van den Berg, J.H. Veldink, M.B. Harms, J.D. Glass, D.J. Stone, P. Tienari, V. Silani, A. Chio, C.E. Shaw, B.J. Traynor and J.E. Landers, *Genome-wide Analyses Identify KIF5A as a Novel ALS Gene*. *Neuron*, 2018. **97**(6): p. 1268-1283 e6.
153. Olahova, M., W.H. Yoon, K. Thompson, S. Jangam, L. Fernandez, J.M. Davidson, J.E. Kyle, M.E. Grove, D.G. Fisk, J.N. Kohler, M. Holmes, A.M. Dries, Y. Huang, C. Zhao, K. Contrepois, Z. Zappala, L. Fresard, D. Waggott, E.M. Zink, Y.M. Kim, H.M. Heyman, K.G. Stratton, B.M. Webb-Robertson, N. Undiagnosed Diseases, M. Snyder, J.D. Merker, S.B. Montgomery, P.G. Fisher, R.G. Feichtinger, J.A. Mayr, J. Hall, I.A. Barbosa, M.A. Simpson, C. Deshpande, K.M. Waters, D.M. Koeller, T.O. Metz, A.A. Morris, S. Schelley, T. Cowan, M.W. Friederich, R. McFarland, J.L.K. Van Hove, G.M. Enns, S. Yamamoto, E.A. Ashley, M.F. Wangler, R.W. Taylor, H.J. Bellen, J.A. Bernstein, and M.T. Wheeler, *Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder*. *Am J Hum Genet*, 2018. **102**(3): p. 494-504.
154. Papanek, B., K.B. O'Dell, P. Manga, R.J. Giannone, D.M. Klingeman, R.L. Hettich, S.D. Brown, and A.M. Guss, *Transcriptomic and proteomic changes from medium supplementation and strain evolution in high-yielding Clostridium thermocellum strains*. *J Ind Microbiol Biotechnol*, 2018.
155. Pena, L.D.M., Y.H. Jiang, K. Schoch, R.C. Spillmann, N. Walley, N. Stong, S. Rapisardo Horn, J.A. Sullivan, A. McConkie-Rosell, S. Kansagra, E.C. Smith, M. El-Dairi, J. Bellet, M.A. Keels, J. Jasien, P.G. Kranz, R. Noel, S.K. Nagaraj, R.K. Lark, D.S.G. Wechsler, D. Del Gaudio, M.L. Leung, L.G. Hendon, C.C. Parker, K.L. Jones, M. Undiagnosed Diseases Network, D.B. Goldstein, and V. Shashi, *Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases*. *Genet Med*, 2018. **20**(4): p. 464-469.
156. Petersen, C.P., A.R. Meyer, C. De Salvo, E. Choi, C. Schlegel, A. Petersen, A.C. Engevik, N. Prasad, S.E. Levy, R.S. Peebles, T.T. Pizarro, and J.R. Goldenring, *A signalling cascade of IL-33 to IL-13 regulates metaplasia in the mouse stomach*. *Gut*, 2018. **67**(5): p. 805-817.

157. Potts, A.H., Y. Leng, P. Babitzke, and T. Romeo, *Examination of Csr regulatory circuitry using epistasis analysis with RNA-seq (Epi-seq) confirms that CsrD affects gene expression via CsrA, CsrB and CsrC*. Sci Rep, 2018. **8**(1): p. 5373.
158. Roberts, B.S., A.A. Hardigan, D.E. Moore, R.C. Ramaker, A.L. Jones, M.B. Fitz-Gerald, G.M. Cooper, C.M. Wilcox, R.P. Kimberly, and R.M. Myers, *Discovery and Validation of Circulating Biomarkers of Colorectal Adenoma by High-Depth Small RNA Sequencing*. Clin Cancer Res, 2018. **24**(9): p. 2092-2099.
159. Schlegel, C., L.A. Lapierre, V.G. Weis, J.A. Williams, I. Kaji, C. Pinzon-Guzman, N. Prasad, B. Boone, A. Jones, H. Correa, S.E. Levy, X. Han, M. Wang, K. Thomsen, S. Acra, and J.R. Goldenring, *Reversible deficits in apical transporter trafficking associated with deficiency in diacylglycerol acyltransferase*. Traffic, 2018.
160. Selvan, N., S. George, F.J. Serajee, M. Shaw, L. Hobson, V. Kalscheuer, N. Prasad, S.E. Levy, J. Taylor, S. Aftimos, C.E. Schwartz, A.M. Huq, J. Gecz, and L. Wells, *O-GlcNAc transferase missense mutations linked to X-linked intellectual disability deregulate genes involved in cell fate determination and signaling*. J Biol Chem, 2018. **293**(27): p. 10810-10824.
161. Shashi, V., K. Schoch, R. Spillmann, H. Cope, Q.K. Tan, N. Walley, L. Pena, A. McConkie-Rosell, Y.H. Jiang, N. Stong, A.C. Need, D.B. Goldstein, and N. Undiagnosed Diseases, *A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative*. Genet Med, 2018.
162. Splinter, K., D.R. Adams, C.A. Bacino, H.J. Bellen, J.A. Bernstein, A.M. Cheattle-Jarvela, C.M. Eng, C. Esteves, W.A. Gahl, R. Hamid, H.J. Jacob, B. Kikani, D.M. Koeller, I.S. Kohane, B.H. Lee, J. Loscalzo, X. Luo, A.T. McCray, T.O. Metz, J.J. Mulvihill, S.F. Nelson, C.G.S. Palmer, J.A. Phillips, 3rd, L. Pick, J.H. Postlethwait, C. Reuter, V. Shashi, D.A. Sweetser, C.J. Tift, N.M. Walley, M.F. Wangler, M. Westerfield, M.T. Wheeler, A.L. Wise, E.A. Worthey, S. Yamamoto, E.A. Ashley, and N. Undiagnosed Diseases, *Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease*. N Engl J Med, 2018. **379**(22): p. 2131-2139.
163. Thompson, M.L., C.R. Finnila, K.M. Bowling, K.B. Brothers, M.B. Neu, M.D. Amaral, S.M. Hiatt, K.M. East, D.E. Gray, J.M.J. Lawlor, W.V. Kelley, E.J. Lose, C.A. Rich, S. Simmons, S.E. Levy, R.M. Myers, G.S. Barsh, E.M. Bebin, and G.M. Cooper, *Genomic sequencing identifies secondary findings in a cohort of parent study participants*. Genet Med, 2018.
164. Walley, N.M., L.D.M. Pena, S.R. Hooper, H. Cope, Y.H. Jiang, A. McConkie-Rosell, C. Sanders, K. Schoch, R.C. Spillmann, K. Strong, A.T. McCray, P. Mazur, C. Esteves, K. LeBlanc, N. Undiagnosed Diseases, A.L. Wise, and V. Shashi, *Characteristics of undiagnosed diseases network applicants: implications for referring providers*. BMC Health Serv Res, 2018. **18**(1): p. 652.
165. Wang, Z., C.L. Wilson, J. Easton, A. Thrasher, H. Mulder, Q. Liu, D.J. Hedges, S. Wang, M.C. Rusch, M.N. Edmonson, S. Levy, J.Q. Lanctot, E. Caron, K. Shelton, K. Currie, M. Lear, A. Patel, C. Rosencrance, Y. Shao, B. Vadodaria, D. Yergeau, Y. Sapkota, R.J. Brooke, W. Moon, E. Rampersaud, X. Ma, T.C. Chang, S.V. Rice, C. Pepper, X. Zhou, X. Chen, W. Chen, A. Jones, B. Boone, M.J. Ehrhardt, M.J. Krasin, R.M. Howell, N.S. Phillips, C. Lewis, D. Srivastava, C.H. Pui, C.A. Kesserwan, G. Wu, K.E. Nichols, J.R. Downing, M.M. Hudson, Y. Yasui, L.L. Robison, and J. Zhang, *Genetic Risk for Subsequent Neoplasms Among Long-Term Survivors of Childhood Cancer*. J Clin Oncol, 2018: p. JCO2018778589.
166. Weiss, H., V.S. Hertzberg, C. Dupont, J.L. Espinoza, S. Levy, K. Nelson, S. Norris, and T. FlyHealthy Research, *The Airplane Cabin Microbiome*. Microb Ecol, 2018.
167. Bhatia, A., B.C. Mobley, J. Cogan, M.E. Koziura, E. Brokamp, J. Phillips, J. Newman, N. Undiagnosed Diseases, S.A. Moore, R. Hamid, and N. Members of the Undiagnosed

- Diseases, *Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy*. Clin Imaging, 2019. **58**: p. 108-113.
168. Burrage, L.C., J.J. Reynolds, N.V. Baratang, J.B. Phillips, J. Wegner, A. McFarquhar, M.R. Higgs, A.E. Christiansen, D.G. Lanza, J.R. Seavitt, M. Jain, X. Li, D.A. Parry, V. Raman, D. Chitayat, I.K. Chinn, A.A. Bertuch, L. Karaviti, A.E. Schlesinger, D. Earl, M. Bamshad, R. Savarirayan, H. Doddapaneni, D. Muzny, S.N. Jhangiani, C.M. Eng, R.A. Gibbs, W. Bi, L. Emrick, J.A. Rosenfeld, J. Postlethwait, M. Westerfield, M.E. Dickinson, A.L. Beaudet, E. Ranza, C. Huber, V. Cormier-Daire, W. Shen, R. Mao, J.D. Heaney, J.S. Orange, G. University of Washington Center for Mendelian, N. Undiagnosed Diseases, D. Bertola, G.L. Yamamoto, W.A.R. Baratela, M.G. Butler, A. Ali, M. Adeli, D.H. Cohn, D. Krakow, A.P. Jackson, M. Lees, A.C. Offiah, C.M. Carlston, J.C. Carey, G.S. Stewart, C.A. Bacino, P.M. Campeau, and B. Lee, *Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes*. Am J Hum Genet, 2019. **104**(3): p. 422-438.
 169. Das, S., R.N.A.C.C. Extracellular, K.M. Ansel, M. Bitzer, X.O. Breakefield, A. Charest, D.J. Galas, M.B. Gerstein, M. Gupta, A. Milosavljevic, M.T. McManus, T. Patel, R.L. Raffai, J. Rozowsky, M.E. Roth, J.A. Saugstad, K. Van Keuren-Jensen, A.M. Weaver, and L.C. Laurent, *The Extracellular RNA Communication Consortium: Establishing Foundational Knowledge and Technologies for Extracellular RNA Research*. Cell, 2019. **177**(2): p. 231-242.
 170. Drange, O.K., O.B. Smeland, A.A. Shadrin, P.I. Finseth, A. Witoelar, O. Frei, G. Psychiatric Genomics Consortium Bipolar Disorder Working, Y. Wang, S. Hassani, S. Djurovic, A.M. Dale, and O.A. Andreassen, *Genetic Overlap Between Alzheimer's Disease and Bipolar Disorder Implicates the MARK2 and VAC14 Genes*. Front Neurosci, 2019. **13**: p. 220.
 171. Grove, M.E., S. White, D.G. Fisk, S. Rego, O. Dagan-Rosenfeld, J.N. Kohler, C.M. Reuter, D. Bonner, N. Undiagnosed Diseases, M.T. Wheeler, J.A. Bernstein, K.E. Ormond, and A.K. Hanson-Kahn, *Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students*. J Genet Couns, 2019. **28**(2): p. 466-476.
 172. Haliyur, R., X. Tong, M. Sanyoura, S. Shrestha, J. Lindner, D.C. Saunders, R. Aramandla, G. Poffenberger, S.D. Redick, R. Bottino, N. Prasad, S.E. Levy, R.D. Blind, D.M. Harlan, L.H. Philipson, R.W. Stein, M. Brissova, and A.C. Powers, *Human islets expressing HNF1A variant have defective beta cell transcriptional regulatory networks*. J Clin Invest, 2019. **129**(1): p. 246-251.
 173. Howard, L.M., J.B. Goll, T.L. Jensen, K.L. Hoek, N. Prasad, C.E. Gelber, S.E. Levy, S. Joyce, A.J. Link, C.B. Creech, and K.M. Edwards, *AS03-Adjuvanted H5N1 Avian Influenza Vaccine Modulates Early Innate Immune Signatures in Human Peripheral Blood Mononuclear Cells*. J Infect Dis, 2019. **219**(11): p. 1786-1798.
 174. Kanca, O., J.C. Andrews, P.T. Lee, C. Patel, S.R. Braddock, A.M. Slavotinek, J.S. Cohen, C.S. Gubbels, K.A. Aldinger, J. Williams, M. Indaram, A. Fatemi, T.W. Yu, P.B. Agrawal, G. Vezina, C. Simons, J. Crawford, C.C. Lau, N. Undiagnosed Diseases, W.K. Chung, T.C. Markello, W.B. Dobyns, D.R. Adams, W.A. Gahl, M.F. Wangler, S. Yamamoto, H.J. Bellen, and M.C.V. Malicdan, *De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia*. Am J Hum Genet, 2019. **105**(3): p. 672-674.
 175. Kelly, M., M. Park, I. Mihalek, A. Roctus, M. Gramm, E. Perez-Palma, E.T. Axeen, C.Y. Hung, H. Olson, L. Swanson, I. Anselm, L.C. Briere, F.A. High, D.A. Sweetser, N. Undiagnosed Diseases, S. Kayani, M. Snyder, S. Calvert, I.E. Scheffer, E. Yang, J.L. Waugh, D. Lal, O. Bodamer, and A. Poduri, *Spectrum of neurodevelopmental disease*

- associated with the GNAO1 guanosine triphosphate-binding region. Epilepsia*, 2019. **60**(3): p. 406-418.
176. Levy, S.E. and B.E. Boone, *Next-Generation Sequencing Strategies*. Cold Spring Harb Perspect Med, 2019. **9**(7).
 177. Ma, X., Y. Shao, L. Tian, D.A. Flasch, H.L. Mulder, M.N. Edmonson, Y. Liu, X. Chen, S. Newman, J. Nakitandwe, Y. Li, B. Li, S. Shen, Z. Wang, S. Shurtleff, L.L. Robison, S. Levy, J. Easton, and J. Zhang, *Analysis of error profiles in deep next-generation sequencing data*. Genome Biol, 2019. **20**(1): p. 50.
 178. Machol, K., J. Rousseau, S. Ehresmann, T. Garcia, T.T.M. Nguyen, R.C. Spillmann, J.A. Sullivan, V. Shashi, Y.H. Jiang, N. Stong, E. Fiala, M. Willing, R. Pfundt, T. Kleefstra, M.T. Cho, H. McLaughlin, M. Rosello Piera, C. Orellana, F. Martinez, A. Caro-Llopis, S. Monfort, T. Roscioli, C.Y. Nixon, M.F. Buckley, A. Turner, W.D. Jones, P.M. van Hasselt, F.C. Hofstede, K.L.I. van Gassen, A.S. Brooks, M.A. van Slegtenhorst, K. Lachlan, J. Sebastian, S. Madan-Khetarpal, D. Sonal, N. Sakkubai, J. Thevenon, L. Faivre, A. Maurel, S. Petrovski, I.D. Krantz, J.M. Tarpinian, J.A. Rosenfeld, B.H. Lee, N. Undiagnosed Diseases, and P.M. Campeau, *Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay*. Am J Hum Genet, 2019. **104**(1): p. 164-178.
 179. McIntyre, A.B.R., R. Ounit, E. Afshinnkoo, R.J. Prill, E. Henaff, N. Alexander, S.S. Minot, D. Danko, J. Foox, S. Ahsanuddin, S. Tighe, N.A. Hasan, P. Subramanian, K. Moffat, S. Levy, S. Lonardi, N. Greenfield, R.R. Colwell, G.L. Rosen, and C.E. Mason, *Correction to: Comprehensive benchmarking and ensemble approaches for metagenomic classifiers*. Genome Biol, 2019. **20**(1): p. 72.
 180. Nicoli, E.R., M.R. Weston, M. Hackbarth, A. Becerril, A. Larson, W.M. Zein, P.R. Baker, 2nd, J.D. Burke, H. Dorward, M. Davids, Y. Huang, D.R. Adams, P.M. Zerfas, D. Chen, T.C. Markello, C. Toro, T. Wood, G. Elliott, M. Vu, N. Undiagnosed Diseases, W. Zheng, L.J. Garrett, C.J. Tiff, W.A. Gahl, D.L. Day-Salvatore, J.A. Mindell, and M.C.V. Malicdan, *Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification*. Am J Hum Genet, 2019. **104**(6): p. 1127-1138.
 181. Panea, R.I., C.L. Love, J.R. Shingleton, A. Reddy, J.A. Bailey, A.M. Moormann, J.A. Otieno, J.M. Ong'echa, C.I. Oduor, K.M.S. Schroeder, N. Masalu, N.J. Chao, M. Agajanian, M.B. Major, Y. Fedoriw, K.L. Richards, G. Rymkiewicz, R.R. Miles, B. Alobeid, G. Bhagat, C.R. Flowers, S.L. Ondrejka, E.D. Hsi, W.W.L. Choi, R.K.H. Au-Yeung, W. Hartmann, G. Lenz, H. Meyerson, Y.Y. Lin, Y. Zhuang, M.A. Luftig, A. Waldrop, T. Dave, D. Thakkar, H. Sahay, G. Li, B.C. Palus, V. Seshadri, S.Y. Kim, R.D. Gascoyne, S. Levy, M. Mukhopadhyay, D.B. Dunson, and S.S. Dave, *The whole-genome landscape of Burkitt lymphoma subtypes*. Blood, 2019. **134**(19): p. 1598-1607.
 182. Saunders, D.C., M. Brissova, N. Phillips, S. Shrestha, J.T. Walker, R. Aramandla, G. Poffenberger, D.K. Flaherty, K.P. Weller, J. Pelletier, T. Cooper, M.T. Goff, J. Virostko, A. Shostak, E.D. Dean, D.L. Greiner, L.D. Shultz, N. Prasad, S.E. Levy, R.H. Carnahan, C. Dai, J. Seigny, and A.C. Powers, *Ectonucleoside Triphosphate Diphosphohydrolase-3 Antibody Targets Adult Human Pancreatic beta Cells for In Vitro and In Vivo Analysis*. Cell Metab, 2019. **29**(3): p. 745-754 e4.
 183. Shashi, V., J. Geist, Y. Lee, Y. Yoo, U. Shin, K. Schoch, J. Sullivan, N. Stong, E. Smith, J. Jasien, P. Kranz, N. Undiagnosed Diseases, Y. Lee, Y.B. Shin, N.T. Wright, M. Choi, and A. Kontogianni-Konstantopoulos, *Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis*. Hum Mutat, 2019. **40**(8): p. 1115-1126.
 184. Stahl, E.A., G. Breen, A.J. Forstner, A. McQuillin, S. Ripke, V. Trubetskoy, M. Mattheisen, Y. Wang, J.R.I. Coleman, H.A. Gaspar, C.A. de Leeuw, S. Steinberg, J.M.W. Pavlides, M. Trzaskowski, E.M. Byrne, T.H. Pers, P.A. Holmans, A.L. Richards, L. Abbott, E. Agerbo,

- H. Akil, D. Albani, N. Alliey-Rodriguez, T.D. Als, A. Anjorin, V. Antilla, S. Awasthi, J.A. Badner, M. Baekvad-Hansen, J.D. Barchas, N. Bass, M. Bauer, R. Belliveau, S.E. Bergen, C.B. Pedersen, E. Boen, M.P. Boks, J. Boocock, M. Budde, W. Bunney, M. Burmeister, J. Bybjerg-Grauholm, W. Byerley, M. Casas, F. Cerrato, P. Cervantes, K. Chambert, A.W. Charney, D. Chen, C. Churchhouse, T.K. Clarke, W. Coryell, D.W. Craig, C. Cruceanu, D. Curtis, P.M. Czerski, A.M. Dale, S. de Jong, F. Degenhardt, J. Del-Favero, J.R. DePaulo, S. Djurovic, A.L. Dobbyn, A. Dumont, T. Elvsashagen, V. Escott-Price, C.C. Fan, S.B. Fischer, M. Flickinger, T.M. Foroud, L. Forty, J. Frank, C. Fraser, N.B. Freimer, L. Frisen, K. Gade, D. Gage, J. Garnham, C. Giambartolomei, M.G. Pedersen, J. Goldstein, S.D. Gordon, K. Gordon-Smith, E.K. Green, M.J. Green, T.A. Greenwood, J. Grove, W. Guan, J. Guzman-Parra, M.L. Hamshire, M. Hautzinger, U. Heilbronner, S. Herms, M. Hipolito, P. Hoffmann, D. Holland, L. Huckins, S. Jamain, J.S. Johnson, A. Jureus, R. Kandaswamy, R. Karlsson, J.L. Kennedy, S. Kittel-Schneider, J.A. Knowles, M. Kogevinas, A.C. Koller, R. Kupka, C. Lavebratt, J. Lawrence, W.B. Lawson, M. Leber, P.H. Lee, S.E. Levy, J.Z. Li, C. Liu, S. Lucae, A. Maaser, D.J. MacIntyre, P.B. Mahon, W. Maier, L. Martinsson, S. McCarroll, P. McGuffin, M.G. McInnis, J.D. McKay, H. Medeiros, S.E. Medland, F. Meng, L. Milani, G.W. Montgomery, D.W. Morris, T.W. Muhleisen, N. Mullins, H. Nguyen, C.M. Nievergelt, A.N. Adolfsson, E.A. Nwulia, C. O'Donovan, L.M.O. Loohuis, A.P.S. Ori, L. Oruc, U. Osby, R.H. Perlis, A. Perry, A. Pfennig, J.B. Potash, S.M. Purcell, E.J. Regeer, A. Reif, C.S. Reinbold, J.P. Rice, F. Rivas, M. Rivera, P. Roussos, D.M. Ruderfer, E. Ryu, C. Sanchez-Mora, A.F. Schatzberg, W.A. Scheftner, N.J. Schork, C. Shannon Weickert, T. Shekhtman, P.D. Shilling, E. Sigurdsson, C. Slaney, O.B. Smeland, J.L. Sobell, C. Soholm Hansen, A.T. Spijker, D. St Clair, M. Steffens, J.S. Strauss, F. Streit, J. Strohmaier, S. Szlinger, R.C. Thompson, T.E. Thorgeirsson, J. Treutlein, H. Vedder, W. Wang, S.J. Watson, T.W. Weickert, S.H. Witt, S. Xi, W. Xu, A.H. Young, P. Zandi, P. Zhang, S. Zollner, Q.C. e, B. Consortium, R. Adolfsson, I. Agartz, M. Alda, L. Backlund, B.T. Baune, F. Bellivier, W.H. Berrettini, J.M. Biernacka, D.H.R. Blackwood, M. Boehnke, A.D. Borglum, A. Corvin, N. Craddock, M.J. Daly, U. Dannlowski, T. Esko, B. Etain, M. Frye, J.M. Fullerton, E.S. Gershon, M. Gill, F. Goes, M. Grigoriu-Serbanescu, J. Hauser, D.M. Hougaard, C.M. Hultman, I. Jones, L.A. Jones, R.S. Kahn, G. Kirov, M. Landen, M. Leboyer, C.M. Lewis, Q.S. Li, J. Lissowska, N.G. Martin, F. Mayoral, S.L. McElroy, A.M. McIntosh, F.J. McMahon, I. Melle, A. Metspalu, P.B. Mitchell, G. Morken, O. Mors, P.B. Mortensen, B. Muller-Myhsok, R.M. Myers, B.M. Neale, V. Nimgaonkar, M. Nordentoft, M.M. Nothen, M.C. O'Donovan, K.J. Oedegaard, M.J. Owen, S.A. Paciga, C. Pato, M.T. Pato, D. Posthuma, J.A. Ramos-Quiroga, M. Ribases, M. Rietschel, G.A. Rouleau, M. Schalling, P.R. Schofield, T.G. Schulze, A. Serretti, J.W. Smoller, H. Stefansson, K. Stefansson, E. Stordal, P.F. Sullivan, G. Turecki, A.E. Vaaler, E. Vieta, J.B. Vincent, T. Werge, J.I. Nurnberger, N.R. Wray, A. Di Florio, H.J. Edenberg, S. Cichon, R.A. Ophoff, L.J. Scott, O.A. Andreassen, J. Kelsoe, P. Sklar and C. Bipolar Disorder Working Group of the Psychiatric Genomics, *Genome-wide association study identifies 30 loci associated with bipolar disorder*. Nat Genet, 2019. **51**(5): p. 793-803.
185. Wilson, C.L., Z. Wang, Q. Liu, M.J. Ehrhardt, R. Mostafavi, J. Easton, H. Mulder, D.J. Hedges, S. Wang, M. Rusch, M. Edmonson, S. Levy, J.Q. Lanctot, K. Currie, M. Lear, A. Patel, Y. Sapkota, R.J. Brooke, W. Moon, T.C. Chang, W. Chen, C.A. Kesserwan, G. Wu, K.E. Nichols, M.M. Hudson, J. Zhang, L.L. Robison, and Y. Yasui, *Estimated number of adult survivors of childhood cancer in United States with cancer-predisposing germline variants*. Pediatr Blood Cancer, 2019: p. e28047.
186. Zastrow, D.B., J.N. Kohler, D. Bonner, C.M. Reuter, L. Fernandez, M.E. Grove, D.G. Fisk, N. Undiagnosed Diseases, Y. Yang, C.M. Eng, P.A. Ward, D. Bick, E.A. Worthey, P.G. Fisher, E.A. Ashley, J.A. Bernstein, and M.T. Wheeler, 3rd, *A toolkit for genetics*

- providers in follow-up of patients with non-diagnostic exome sequencing.* J Genet Couns, 2019. **28**(2): p. 213-228.
187. Butler, D.J., C. Mozsary, C. Meydan, D. Danko, J. Foox, J. Rosiene, A. Shaiber, E. Afshinnekoo, M. MacKay, F.J. Sedlazeck, N.A. Ivanov, M. Sierra, D. Pohle, M. Zietz, U. Gisladdottir, V. Ramlall, C.D. Westover, K. Ryon, B. Young, C. Bhattacharya, P. Ruggiero, B.W. Langhorst, N. Tanner, J. Gawrys, D. Meleshko, D. Xu, P.A.D. Steel, A.J. Shemesh, J. Xiang, J. Thierry-Mieg, D. Thierry-Mieg, R.E. Schwartz, A. Iftner, D. Bezdan, J. Siple, L. Cong, A. Craney, P. Velu, A.M. Melnick, I. Hajirasouliha, S.M. Horner, T. Iftner, M. Salvatore, M. Loda, L.F. Westblade, M. Cushing, S. Levy, S. Wu, N. Tatonetti, M. Imielinski, H. Rennert, and C.E. Mason, *Shotgun Transcriptome and Isothermal Profiling of SARS-CoV-2 Infection Reveals Unique Host Responses, Viral Diversification, and Drug Interactions.* bioRxiv, 2020.
 188. Coleman, J.R.I., H.A. Gaspar, J. Bryois, C. Bipolar Disorder Working Group of the Psychiatric Genomics, C. Major Depressive Disorder Working Group of the Psychiatric Genomics, and G. Breen, *The Genetics of the Mood Disorder Spectrum: Genome-wide Association Analyses of More Than 185,000 Cases and 439,000 Controls.* Biol Psychiatry, 2020. **88**(2): p. 169-184.
 189. Consortium, E.P., M.P. Snyder, T.R. Gingeras, J.E. Moore, Z. Weng, M.B. Gerstein, B. Ren, R.C. Hardison, J.A. Stamatoyannopoulos, B.R. Graveley, E.A. Feingold, M.J. Pazin, M. Pagan, D.A. Gilchrist, B.C. Hitz, J.M. Cherry, B.E. Bernstein, E.M. Mendenhall, D.R. Zerbino, A. Frankish, P. Flicek, and R.M. Myers, *Perspectives on ENCODE.* Nature, 2020. **583**(7818): p. 693-698.
 190. Dai, C., J.T. Walker, A. Shostak, A. Padgett, E. Spears, S. Wisniewski, G. Poffenberger, R. Aramandla, E.D. Dean, N. Prasad, S.E. Levy, D.L. Greiner, L.D. Shultz, R. Bottino, and A.C. Powers, *Tacrolimus- and sirolimus-induced human beta cell dysfunction is reversible and preventable.* JCI Insight, 2020. **5**(1).
 191. Johnson, B.V., R. Kumar, S. Oishi, S. Alexander, M. Kasherman, M.S. Vega, A. Ivancevic, A. Gardner, D. Domingo, M. Corbett, E. Parnell, S. Yoon, T. Oh, M. Lines, H. Lefroy, U. Kini, M. Van Allen, S. Gronborg, S. Mercier, S. Kury, S. Bezieau, L. Pasquier, M. Raynaud, A. Afenjar, T. Billette de Villemeur, B. Keren, J. Desir, L. Van Maldergem, M. Marangoni, N. Dikow, D.A. Koolen, P.M. VanHasselt, M. Weiss, P. Zwijnenburg, J. Sa, C.F. Reis, C. Lopez-Otin, O. Santiago-Fernandez, A. Fernandez-Jaen, A. Rauch, K. Steindl, P. Joset, A. Goldstein, S. Madan-Khetarpal, E. Infante, E. Zackai, C. McDougall, V. Narayanan, K. Ramsey, S. Mercimek-Andrews, L. Pena, V. Shashi, N. Undiagnosed Diseases, K. Schoch, J.A. Sullivan, E.V.F. Pinto, P.N. Pichurin, S.A. Ewing, S.S. Barnett, E.W. Klee, M.S. Perry, M.K. Koenig, C.E. Keegan, J.L. Schuette, S. Asher, Y. Perilla-Young, L.D. Smith, J.A. Rosenfeld, E. Bhoj, P. Kaplan, D. Li, R. Oegema, E. van Binsbergen, B. van der Zwaag, M.F. Smeland, I. Cutcutache, M. Page, M. Armstrong, A.E. Lin, M.A. Steeves, N.D. Hollander, M.J.V. Hoffer, M.R.F. Reijnders, S. Demirdas, D.C. Koboldt, D. Bartholomew, T.M. Mosher, S.E. Hickey, C. Shieh, P.A. Sanchez-Lara, J.M. Graham, Jr., K. Tezcan, G.B. Schaefer, N.R. Danylchuk, A. Asamoah, K.E. Jackson, N. Yachelevich, M. Au, L.A. Perez-Jurado, T. Kleefstra, P. Penzes, S.A. Wood, T. Burne, T.M. Pierson, M. Piper, J. Gecz and L.A. Jolly, *Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor beta Signaling.* Biol Psychiatry, 2020. **87**(2): p. 100-112.
 192. Koehler, J., M. Sandey, N. Prasad, S.A. Levy, X. Wang, and X. Wang, *Differential Expression of miRNAs in Hypoxia ("HypoxamiRs") in Three Canine High-Grade Glioma Cell Lines.* Front Vet Sci, 2020. **7**: p. 104.
 193. MacKay, M.J., A.C. Hooker, E. Afshinnekoo, M. Salit, J. Kelly, J.V. Feldstein, N. Haft, D. Schenkel, S. Nambi, Y. Cai, F. Zhang, G. Church, J. Dai, C.L. Wang, S. Levy, J. Huber,

- H.P. Ji, A. Kriegel, A.L. Wyllie, and C.E. Mason, *The COVID-19 XPRIZE and the need for scalable, fast, and widespread testing*. Nat Biotechnol, 2020. **38**(9): p. 1021-1024.
194. Rowneki, M., N. Aronson, P. Du, P. Sachs, R. Blakemore, S. Chakravorty, S. Levy, A.L. Jones, G. Trivedi, S. Chebore, D. Addo, D.K. Byarugaba, P.D. Njobvu, F. Wabwire-Mangen, B. Erima, E.S. Ramos, C.A. Evans, B. Hale, J.D. Mancuso, and D. Alland, *Detection of drug resistant Mycobacterium tuberculosis by high-throughput sequencing of DNA isolated from acid fast bacilli smears*. PLoS One, 2020. **15**(5): p. e0232343.
195. Svrzikapa, N., K.A. Longo, N. Prasad, R. Boyanapalli, J.M. Brown, D. Dorset, S. Yourstone, J. Powers, S.E. Levy, A.J. Morris, C. Vargeese, and J. Goyal, *Investigational Assay for Haplotype Phasing of the Huntingtin Gene*. Mol Ther Methods Clin Dev, 2020. **19**: p. 162-173.
196. Tomoiaga, D., V. Aguiar-Pulido, S. Shrestha, P. Feinstein, S.E. Levy, C.E. Mason, and J.A. Rosenfeld, *Single-cell sperm transcriptomes and variants from fathers of children with and without autism spectrum disorder*. NPJ Genom Med, 2020. **5**: p. 14.
197. Wilson, C.L., Z. Wang, Q. Liu, M.J. Ehrhardt, R. Mostafavi, J. Easton, H. Mulder, D.J. Hedges, S. Wang, M. Rusch, M. Edmonson, S. Levy, J.Q. Lanctot, K. Currie, M. Lear, A. Patel, Y. Sapkota, R.J. Brooke, W. Moon, T.C. Chang, W. Chen, C.A. Kesserwan, G. Wu, K.E. Nichols, M.M. Hudson, J. Zhang, L.L. Robison, and Y. Yasui, *Estimated number of adult survivors of childhood cancer in United States with cancer-predisposing germline variants*. Pediatr Blood Cancer, 2020. **67**(2): p. e28047.
198. Agarwal, P., M.P. Crepps, N.A. Stahr, W.P. Kretzschmar, H.C. Harris, N. Prasad, S.E. Levy, and B.F. Smith, *Identification of canine circulating miRNAs as tumor biospecific markers using Next-Generation Sequencing and Q-RT-PCR*. Biochem Biophys Rep, 2021. **28**: p. 101106.
199. Andlauer, T.F.M., J. Guzman-Parra, F. Streit, J. Strohmaier, M.J. Gonzalez, S. Gil Flores, F.J. Cabaleiro Fabeiro, F. Del Rio Noriega, F.P. Perez, J. Haro Gonzalez, G. Orozco Diaz, Y. de Diego-Otero, B. Moreno-Kustner, G. Auburger, F. Degenhardt, S. Heilmann-Heimbach, S. Herms, P. Hoffmann, J. Frank, J.C. Foo, J. Treutlein, S.H. Witt, S. Cichon, M. Kogevinas, C. Bipolar Disorder Working Group of the Psychiatric Genomics, C. Major Depressive Disorder Working Group of the Psychiatric Genomics, F. Rivas, F. Mayoral, B. Muller-Myhsok, A.J. Forstner, M.M. Nothen, and M. Rietschel, *Bipolar multiplex families have an increased burden of common risk variants for psychiatric disorders*. Mol Psychiatry, 2021. **26**(4): p. 1286-1298.
200. Brokamp, E., M.E. Koziura, J.A. Phillips, 3rd, L.A. Tang, J.D. Cogan, L.C. Rives, A.K. Robertson, L. Duncan, A. Bican, J.F. Peterson, J.H. Newman, R. Hamid, L. Bastarache, and N. Undiagnosed Diseases, *One is the loneliest number: genotypic matchmaking using the electronic health record*. Genet Med, 2021. **23**(10): p. 1830-1832.
201. Butler, D., C. Mozsary, C. Meydan, J. Foox, J. Rosiene, A. Shaiber, D. Danko, E. Afshinnikoo, M. MacKay, F.J. Sedlazeck, N.A. Ivanov, M. Sierra, D. Pohle, M. Zietz, U. Gisladdottir, V. Ramlall, E.T. Sholle, E.J. Schenck, C.D. Westover, C. Hassan, K. Ryon, B. Young, C. Bhattacharya, D.L. Ng, A.C. Granados, Y.A. Santos, V. Servellita, S. Federman, P. Ruggiero, A. Fungtammasan, C.S. Chin, N.M. Pearson, B.W. Langhorst, N.A. Tanner, Y. Kim, J.W. Reeves, T.D. Hether, S.E. Warren, M. Bailey, J. Gawrys, D. Meleshko, D. Xu, M. Couto-Rodriguez, D. Nagy-Szakal, J. Barrows, H. Wells, N.B. O'Hara, J.A. Rosenfeld, Y. Chen, P.A.D. Steel, A.J. Shemesh, J. Xiang, J. Thierry-Mieg, D. Thierry-Mieg, A. Iftner, D. Bezdan, E. Sanchez, T.R. Campion, Jr., J. Siple, L. Cong, A. Craney, P. Velu, A.M. Melnick, S. Shapira, I. Hajirasouliha, A. Borczuk, T. Iftner, M. Salvatore, M. Loda, L.F. Westblade, M. Cushing, S. Wu, S. Levy, C. Chiu, R.E. Schwartz, N. Tatonetti, H. Rennert, M. Imielinski, and C.E. Mason, *Shotgun transcriptome, spatial omics, and isothermal profiling of SARS-CoV-2 infection reveals unique host responses, viral diversification, and drug interactions*. Nat Commun, 2021. **12**(1): p. 1660.

202. De La Vega, F.M., S. Chowdhury, B. Moore, E. Frise, J. McCarthy, E.J. Hernandez, T. Wong, K. James, L. Guidugli, P.B. Agrawal, C.A. Genetti, C.A. Brownstein, A.H. Beggs, B.S. Loscher, A. Franke, B. Boone, S.E. Levy, K. Ounap, S. Pajusalu, M. Huentelman, K. Ramsey, M. Naymik, V. Narayanan, N. Veeraraghavan, P. Billings, M.G. Reese, M. Yandell, and S.F. Kingsmore, *Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases*. *Genome Med*, 2021. **13**(1): p. 153.
203. Foox, J., S.W. Tighe, C.M. Nicolet, J.M. Zook, M. Byrska-Bishop, W.E. Clarke, M.M. Khayat, M. Mahmoud, P.K. Laaguiby, Z.T. Herbert, D. Warner, G.S. Grills, J. Jen, S. Levy, J. Xiang, A. Alonso, X. Zhao, W. Zhang, F. Teng, Y. Zhao, H. Lu, G.P. Schroth, G. Narzisi, W. Farmerie, F.J. Sedlazeck, D.A. Baldwin, and C.E. Mason, *Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study*. *Nat Biotechnol*, 2021. **39**(9): p. 1129-1140.
204. Jia, X., F.S. Goes, A.E. Locke, D. Palmer, W. Wang, S. Cohen-Woods, G. Genovese, A.U. Jackson, C. Jiang, M. Kvale, N. Mullins, H. Nguyen, M. Pirooznia, M. Rivera, D.M. Ruderfer, L. Shen, K. Thai, M. Zawistowski, Y. Zhuang, G. Abecasis, H. Akil, S. Bergen, M. Burmeister, S. Chapman, M. DelaBastide, A. Jureus, H.M. Kang, P.Y. Kwok, J.Z. Li, S.E. Levy, E.T. Monson, J. Moran, J. Sobell, S. Watson, V. Willour, S. Zollner, R. Adolfsson, D. Blackwood, M. Boehnke, G. Breen, A. Corvin, N. Craddock, A. DiFlorio, C.M. Hultman, M. Landen, C. Lewis, S.A. McCarroll, W. Richard McCombie, P. McGuffin, A. McIntosh, A. McQuillin, D. Morris, R.M. Myers, M. O'Donovan, R. Ophoff, M. Boks, R. Kahn, W. Ouwehand, M. Owen, C. Pato, M. Pato, D. Posthuma, J.B. Potash, A. Reif, P. Sklar, J. Smoller, P.F. Sullivan, J. Vincent, J. Walters, B. Neale, S. Purcell, N. Risch, C. Schaefer, E.A. Stahl, P.P. Zandi, and L.J. Scott, *Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder*. *Mol Psychiatry*, 2021. **26**(9): p. 5239-5250.
205. Karlebach, G., B. Aronow, S.B. Baylin, D. Butler, J. Foox, S. Levy, C. Meydan, C. Mozsary, A.M. Saravia-Butler, D.M. Taylor, E. Wurtele, C.E. Mason, A. Beheshti, and P.N. Robinson, *Betacoronavirus-specific alternate splicing*. *bioRxiv*, 2021.
206. Keele, G.R., J.W. Prokop, H. He, K. Holl, J. Littrell, A.W. Deal, Y. Kim, P.B. Kyle, E. Attipoe, A.C. Johnson, K.L. Uhl, O.L. Sirpilla, S. Jahanbakhsh, M. Robinson, S. Levy, W. Valdar, M.R. Garrett, and L.C. Solberg Woods, *Sept8/SEPTIN8 involvement in cellular structure and kidney damage is identified by genetic mapping and a novel human tubule hypoxic model*. *Sci Rep*, 2021. **11**(1): p. 2071.
207. Kyle, J.E., K.G. Stratton, E.M. Zink, Y.M. Kim, K.J. Bloodsworth, M.E. Monroe, N. Undiagnosed Diseases, K.M. Waters, B.M. Webb-Robertson, D.M. Koeller, and T.O. Metz, *A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases*. *Sci Data*, 2021. **8**(1): p. 114.
208. Mullins, N., A.J. Forstner, K.S. O'Connell, B. Coombes, J.R.I. Coleman, Z. Qiao, T.D. Als, T.B. Bigdeli, S. Borte, J. Bryois, A.W. Charney, O.K. Drange, M.J. Gandal, S.P. Hagenaars, M. Ikeda, N. Kamitaki, M. Kim, K. Krebs, G. Panagiotaropoulou, B.M. Schilder, L.G. Sloofman, S. Steinberg, V. Trubetskoy, B.S. Winsvold, H.H. Won, L. Abramova, K. Adorjan, E. Agerbo, M. Al Eissa, D. Albani, N. Alliey-Rodriguez, A. Anjorin, V. Antilla, A. Antoniou, S. Awasthi, J.H. Baek, M. Baekvad-Hansen, N. Bass, M. Bauer, E.C. Beins, S.E. Bergen, A. Birner, C. Bocker Pedersen, E. Boen, M.P. Boks, R. Bosch, M. Brum, B.M. Brumpton, N. Brunkhorst-Kanaan, M. Budde, J. Bybjerg-Grauholm, W. Byerley, M. Cairns, M. Casas, P. Cervantes, T.K. Clarke, C. Cruceanu, A. Cuellar-Barboza, J. Cunningham, D. Curtis, P.M. Czerski, A.M. Dale, N. Dalkner, F.S. David, F. Degenhardt, S. Djurovic, A.L. Dobbyn, A. Douzenis, T. Elvsashagen, V. Escott-Price, I.N. Ferrier, A. Fiorentino, T.M. Foroud, L. Forty, J. Frank, O. Frei, N.B. Freimer, L. Frisen, K. Gade, J. Garnham, J. Gelernter, M. Giortz Pedersen, I.R. Gizer, S.D. Gordon, K. Gordon-

- Smith, T.A. Greenwood, J. Grove, J. Guzman-Parra, K. Ha, M. Haraldsson, M. Hautzinger, U. Heilbronner, D. Hellgren, S. Herms, P. Hoffmann, P.A. Holmans, L. Huckins, S. Jamain, J.S. Johnson, J.L. Kalman, Y. Kamatani, J.L. Kennedy, S. Kittel-Schneider, J.A. Knowles, M. Kogevinas, M. Koromina, T.M. Kranz, H.R. Kranzler, M. Kubo, R. Kupka, S.A. Kushner, C. Lavebratt, J. Lawrence, M. Leber, H.J. Lee, P.H. Lee, S.E. Levy, C. Lewis, C. Liao, S. Lucae, M. Lundberg, D.J. MacIntyre, S.H. Magnusson, W. Maier, A. Maihofer, D. Malaspina, E. Maratou, L. Martinsson, M. Mattheisen, S.A. McCarroll, N.W. McGregor, P. McGuffin, J.D. McKay, H. Medeiros, S.E. Medland, V. Millischer, G.W. Montgomery, J.L. Moran, D.W. Morris, T.W. Muhleisen, N. O'Brien, C. O'Donovan, L.M. Olde Loohuis, L. Oruc, S. Papiol, A.F. Pardinas, A. Perry, A. Pfennig, E. Porichi, J.B. Potash, D. Quested, T. Raj, M.H. Rapaport, J.R. DePaulo, E.J. Regeer, J.P. Rice, F. Rivas, M. Rivera, J. Roth, P. Roussos, D.M. Ruderfer, C. Sanchez-Mora, E.C. Schulte, F. Senner, S. Sharp, P.D. Shilling, E. Sigurdsson, L. Sirignano, C. Slaney, O.B. Smeland, D.J. Smith, J.L. Sobell, C. Soholm Hansen, M. Soler Artigas, A.T. Spijker, D.J. Stein, J.S. Strauss, B. Swiatkowska, C. Terao, T.E. Thorgeirsson, C. Toma, P. Tooney, E.E. Tsermpini, M.P. Vawter, H. Vedder, J.T.R. Walters, S.H. Witt, S. Xi, W. Xu, J.M.K. Yang, A.H. Young, H. Young, P.P. Zandi, H. Zhou, L. Zillich, H.A.-I. Psychiatry, R. Adolfsson, I. Agartz, M. Alda, L. Alfredsson, G. Babadjanova, L. Backlund, B.T. Baune, F. Bellivier, S. Bengesser, W.H. Berrettini, D.H.R. Blackwood, M. Boehnke, A.D. Borglum, G. Breen, V.J. Carr, S. Catts, A. Corvin, N. Craddock, U. Dannlowski, D. Dikeos, T. Esko, B. Etain, P. Ferentinos, M. Frye, J.M. Fullerton, M. Gawlik, E.S. Gershon, F.S. Goes, M.J. Green, M. Grigoriou-Serbanescu, J. Hauser, F. Henskens, J. Hillert, K.S. Hong, D.M. Hougaard, C.M. Hultman, K. Hveem, N. Iwata, A.V. Jablensky, I. Jones, L.A. Jones, R.S. Kahn, J.R. Kelsoe, G. Kirov, M. Landen, M. Leboyer, C.M. Lewis, Q.S. Li, J. Lissowska, C. Lochner, C. Loughland, N.G. Martin, C.A. Mathews, F. Mayoral, S.L. McElroy, A.M. McIntosh, F.J. McMahon, I. Melle, P. Michie, L. Milani, P.B. Mitchell, G. Morken, O. Mors, P.B. Mortensen, B. Mowry, B. Muller-Myhsok, R.M. Myers, B.M. Neale, C.M. Nievergelt, M. Nordentoft, M.M. Nothen, M.C. O'Donovan, K.J. Oedegaard, T. Olsson, M.J. Owen, S.A. Paciga, C. Pantelis, C. Pato, M.T. Pato, G.P. Patrinos, R.H. Perlis, D. Posthuma, J.A. Ramos-Quiroga, A. Reif, E.Z. Reininghaus, M. Ribases, M. Rietschel, S. Ripke, G.A. Rouleau, T. Saito, U. Schall, M. Schalling, P.R. Schofield, T.G. Schulze, L.J. Scott, R.J. Scott, A. Serretti, C. Shannon Weickert, J.W. Smoller, H. Stefansson, K. Stefansson, E. Stordal, F. Streit, P.F. Sullivan, G. Turecki, A.E. Vaaler, E. Vieta, J.B. Vincent, I.D. Waldman, T.W. Weickert, T. Werge, N.R. Wray, J.A. Zwart, J.M. Biernacka, J.I. Nurnberger, S. Cichon, H.J. Edenberg, E.A. Stahl, A. McQuillin, A. Di Florio, R.A. Ophoff and O.A. Andreassen, *Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology*. Nat Genet, 2021. **53**(6): p. 817-829.
209. Park, J., J. Foxx, T. Hether, D. Danko, S. Warren, Y. Kim, J. Reeves, D.J. Butler, C. Mozsary, J. Rosiene, A. Shaiber, E. Afshinnekoo, M. MacKay, Y. Bram, V. Chandar, H. Geiger, A. Craney, P. Velu, A.M. Melnick, I. Hajirasouliha, A. Beheshti, D. Taylor, A. Saravia-Butler, U. Singh, E.S. Wurtele, J. Schisler, S. Fennessey, A. Corvelo, M.C. Zody, S. Germer, S. Salvatore, S. Levy, S. Wu, N. Tatonetti, S. Shapira, M. Salvatore, M. Loda, L.F. Westblade, M. Cushing, H. Rennert, A.J. Kriegel, O. Elemento, M. Imielinski, A.C. Borczuk, C. Meydan, R.E. Schwartz, and C.E. Mason, *Systemic Tissue and Cellular Disruption from SARS-CoV-2 Infection revealed in COVID-19 Autopsies and Spatial Omics Tissue Maps*. bioRxiv, 2021.
210. Rampersaud, E., G. Kang, L.E. Palmer, S.R. Rashkin, S. Wang, W. Bi, N.M. Alberts, D. Anghelescu, M. Barton, K. Birch, N. Boulos, A.M. Brandow, R.J. Brooke, T.C. Chang, W. Chen, Y. Cheng, J. Ding, J. Easton, J.R. Hodges, C.K. Kanne, S. Levy, H. Mulder, A.P. Patel, L. Puri, C. Rosencrance, M. Rusch, Y. Sapkota, E. Sioson, A. Sharma, X. Tang, A.

- Thrasher, W. Wang, Y. Yao, Y. Yasui, D. Yergeau, J.S. Hankins, V.A. Sheehan, J.R. Downing, J.H. Estep, J. Zhang, M. DeBaun, G. Wu, and M.J. Weiss, *A polygenic score for acute vaso-occlusive pain in pediatric sickle cell disease*. *Blood Adv*, 2021. **5**(14): p. 2839-2851.
211. Saunders, D.C., K.I. Aamodt, T.M. Richardson, A.J. Hopkirk, R. Aramandla, G. Poffenberger, R. Jenkins, D.K. Flaherty, N. Prasad, S.E. Levy, A.C. Powers, and M. Brissova, *Coordinated interactions between endothelial cells and macrophages in the islet microenvironment promote beta cell regeneration*. *NPJ Regen Med*, 2021. **6**(1): p. 22.
212. Shrestha, S., D.C. Saunders, J.T. Walker, J. Camunas-Soler, X.Q. Dai, R. Haliyur, R. Aramandla, G. Poffenberger, N. Prasad, R. Bottino, R. Stein, J.P. Cartailier, S.C. Parker, P.E. MacDonald, S.E. Levy, A.C. Powers, and M. Brissova, *Combinatorial transcription factor profiles predict mature and functional human islet alpha and beta cells*. *JCI Insight*, 2021. **6**(18).
213. Blokland, G.A.M., J. Grove, C.Y. Chen, C. Cotsapas, S. Tobet, R. Handa, C. Schizophrenia Working Group of the Psychiatric Genomics, D. St Clair, T. Lencz, B.J. Mowry, S. Periyasamy, M.J. Cairns, P.A. Tooney, J.Q. Wu, B. Kelly, G. Kirov, P.F. Sullivan, A. Corvin, B.P. Riley, T. Esko, L. Milani, E.G. Jonsson, A. Palotie, H. Ehrenreich, M. Begemann, A. Steixner-Kumar, P.C. Sham, N. Iwata, D.R. Weinberger, P.V. Gejman, A.R. Sanders, J.D. Buxbaum, D. Rujescu, I. Giegling, B. Konte, A.M. Hartmann, E. Bramon, R.M. Murray, M.T. Pato, J. Lee, I. Melle, E. Molden, R.A. Ophoff, A. McQuillin, N.J. Bass, R. Adolfsson, A.K. Malhotra, C. Bipolar Disorder Working Group of the Psychiatric Genomics, N.G. Martin, J.M. Fullerton, P.B. Mitchell, P.R. Schofield, A.J. Forstner, F. Degenhardt, S. Schaupp, A.L. Comes, M. Kogevinas, J. Guzman-Parra, A. Reif, F. Streit, L. Sirignano, S. Cichon, M. Grigoriu-Serbanescu, J. Hauser, J. Lissowska, F. Mayoral, B. Muller-Myhsok, B. Swiatkowska, T.G. Schulze, M.M. Nothen, M. Rietschel, J. Kelsoe, M. Leboyer, S. Jamain, B. Etain, F. Bellivier, J.B. Vincent, M. Alda, C. O'Donovan, P. Cervantes, J.M. Biernacka, M. Frye, S.L. McElroy, L.J. Scott, E.A. Stahl, M. Landen, M.L. Hamshire, O.B. Smeland, S. Djurovic, A.E. Vaaler, O.A. Andreassen, C. Major Depressive Disorder Working Group of the Psychiatric Genomics, B.T. Baune, T. Air, M. Preisig, R. Uher, D.F. Levinson, M.M. Weissman, J.B. Potash, J. Shi, J.A. Knowles, R.H. Perlis, S. Lucae, D.I. Boomsma, B. Penninx, J.J. Hottenga, E.J.C. de Geus, G. Willemsen, Y. Milaneschi, H. Tiemeier, H.J. Grabe, A. Teumer, S. Van der Auwera, U. Volker, S.P. Hamilton, P.K.E. Magnusson, A. Viktorin, D. Mehta, N. Mullins, M.J. Adams, G. Breen, A.M. McIntosh, C.M. Lewis, C. Sex Differences Cross-Disorder Analysis Group of the Psychiatric Genomics, iPsych, D.M. Hougaard, M. Nordentoft, O. Mors, P.B. Mortensen, T. Werge, T.D. Als, A.D. Borglum, T.L. Petryshen, J.W. Smoller and J.M. Goldstein, *Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders*. *Biol Psychiatry*, 2022. **91**(1): p. 102-117.
214. Karlebach, G., B. Aronow, S.B. Baylin, D. Butler, J. Fook, S. Levy, C. Meydan, C. Mozsary, A.M. Saravia-Butler, D.M. Taylor, E. Wurtele, C.E. Mason, A. Beheshti, and P.N. Robinson, *Betacoronavirus-specific alternate splicing*. *Genomics*, 2022. **114**(2): p. 110270.
215. McClean, P.E., R. Lee, K. Howe, C. Osborne, J. Grimwood, S. Levy, A.P. Haugrud, C. Plott, M. Robinson, R.M. Skiba, T. Tanha, M. Zamani, T.W. Thannhauser, R.P. Glahn, J. Schmutz, J.M. Osorno, and P.N. Miklas, *The Common Bean V Gene Encodes Flavonoid 3'5' Hydroxylase: A Major Mutational Target for Flavonoid Diversity in Angiosperms*. *Front Plant Sci*, 2022. **13**: p. 869582.
216. Mullins, N., J. Kang, A.I. Campos, J.R.I. Coleman, A.C. Edwards, H. Galfalvy, D.F. Levey, A. Lori, A. Shabalín, A. Starnawska, M.H. Su, H.J. Watson, M. Adams, S. Awasthi, M. Gandal, J.D. Hafferty, A. Hishimoto, M. Kim, S. Okazaki, I. Otsuka, S. Ripke, E.B. Ware,

- A.W. Bergen, W.H. Berrettini, M. Bohus, H. Brandt, X. Chang, W.J. Chen, H.C. Chen, S. Crawford, S. Crow, E. DiBlasi, P. Duriez, F. Fernandez-Aranda, M.M. Fichter, S. Gallinger, S.J. Glatt, P. Gorwood, Y. Guo, H. Hakonarson, K.A. Halmi, H.G. Hwu, S. Jain, S. Jamain, S. Jimenez-Murcia, C. Johnson, A.S. Kaplan, W.H. Kaye, P.K. Keel, J.L. Kennedy, K.L. Klump, D. Li, S.C. Liao, K. Lieb, L. Lilenfeld, C.M. Liu, P.J. Magistretti, C.R. Marshall, J.E. Mitchell, E.T. Monson, R.M. Myers, D. Pinto, A. Powers, N. Ramoz, S. Roepke, V. Rozanov, S.W. Scherer, C. Schmah, M. Sokolowski, M. Strober, L.M. Thornton, J. Treasure, M.T. Tsuang, S.H. Witt, D.B. Woodside, Z. Yilmaz, L. Zillich, R. Adolfsson, I. Agartz, T.M. Air, M. Alda, L. Alfredsson, O.A. Andreassen, A. Anjorin, V. Appadurai, M. Soler Artigas, S. Van der Auwera, M.H. Azevedo, N. Bass, C.H.D. Bau, B.T. Baune, F. Bellivier, K. Berger, J.M. Biernacka, T.B. Bigdeli, E.B. Binder, M. Boehnke, M.P. Boks, R. Bosch, D.L. Braff, R. Bryant, M. Budde, E.M. Byrne, W. Cahn, M. Casas, E. Castela, J.A. Cervilla, B. Chaumette, S. Cichon, A. Corvin, N. Craddock, D. Craig, F. Degenhardt, S. Djurovic, H.J. Edenberg, A.H. Fanous, J.C. Foo, A.J. Forstner, M. Frye, J.M. Fullerton, J.M. Gatt, P.V. Gejman, I. Giegling, H.J. Grabe, M.J. Green, E.H. Grevet, M. Grigoriu-Serbanescu, B. Gutierrez, J. Guzman-Parra, S.P. Hamilton, M.L. Hamshire, A. Hartmann, J. Hauser, S. Heilmann-Heimbach, P. Hoffmann, M. Ising, I. Jones, L.A. Jones, L. Jonsson, R.S. Kahn, J.R. Kelsoe, K.S. Kendler, S. Kloiber, K.C. Koenen, M. Kogevinas, B. Konte, M.O. Krebs, M. Landen, J. Lawrence, M. Leboyer, P.H. Lee, D.F. Levinson, C. Liao, J. Lissowska, S. Lucae, F. Mayoral, S.L. McElroy, P. McGrath, P. McGuffin, A. McQuillin, S.E. Medland, D. Mehta, I. Melle, Y. Milaneschi, P.B. Mitchell, E. Molina, G. Morken, P.B. Mortensen, B. Muller-Myhsok, C. Nievergelt, V. Nimgaonkar, M.M. Nothen, M.C. O'Donovan, R.A. Ophoff, M.J. Owen, C. Pato, M.T. Pato, B. Penninx, J. Pimm, G. Pistis, J.B. Potash, R.A. Power, M. Preisig, D. Quested, J.A. Ramos-Quiroga, A. Reif, M. Ribases, V. Richarte, M. Rietschel, M. Rivera, A. Roberts, G. Roberts, G.A. Rouleau, D.L. Rovaris, D. Rujescu, C. Sanchez-Mora, A.R. Sanders, P.R. Schofield, T.G. Schulze, L.J. Scott, A. Serretti, J. Shi, S.I. Shyn, L. Sirignano, P. Sklar, O.B. Smeland, J.W. Smoller, E.J.S. Sonuga-Barke, G. Spalletta, J.S. Strauss, B. Swiatkowska, M. Trzaskowski, G. Turecki, L. Vilar-Ribo, J.B. Vincent, H. Volzke, J.T.R. Walters, C. Shannon Weickert, T.W. Weickert, M.M. Weissman, L.M. Williams, N.R. Wray, C.C. Zai, A.E. Ashley-Koch, J.C. Beckham, E.R. Hauser, M.A. Hauser, N.A. Kimbrel, J.H. Lindquist, B. McMahon, D.W. Oslin, X. Qin, C. Major Depressive Disorder Working Group of the Psychiatric Genomics, C. Bipolar Disorder Working Group of the Psychiatric Genomics, C. Eating Disorders Working Group of the Psychiatric Genomics, C. German Borderline Genomics, M.V.P.S.E. Workgroup, V.A.M.V. Program, E. Agerbo, A.D. Borglum, G. Breen, A. Erlangen, T. Esko, J. Gelernter, D.M. Hougaard, R.C. Kessler, H.R. Kranzler, Q.S. Li, N.G. Martin, A.M. McIntosh, O. Mors, M. Nordentoft, C.M. Olsen, D. Porteous, R.J. Ursano, D. Wasserman, T. Werge, D.C. Whiteman, C.M. Bulik, H. Coon, D. Demontis, A.R. Docherty, P.H. Kuo, C.M. Lewis, J.J. Mann, M.E. Renteria, D.J. Smith, E.A. Stahl, M.B. Stein, F. Streit, V. Willour and D.M. Ruderfer, *Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors*. Biol Psychiatry, 2022. **91**(3): p. 313-327.
217. Park, J., J. Foon, T. Hether, D.C. Danko, S. Warren, Y. Kim, J. Reeves, D.J. Butler, C. Mozsary, J. Rosiene, A. Shaiber, E.E. Afshin, M. MacKay, A.F. Rendeiro, Y. Bram, V. Chandar, H. Geiger, A. Craney, P. Velu, A.M. Melnick, I. Hajirasouliha, A. Beheshti, D. Taylor, A. Saravia-Butler, U. Singh, E.S. Wurtele, J. Schisler, S. Fennessey, A. Corvelo, M.C. Zody, S. Germer, S. Salvatore, S. Levy, S. Wu, N.P. Tatonetti, S. Shapira, M. Salvatore, L.F. Westblade, M. Cushing, H. Rennert, A.J. Kriegel, O. Elemento, M. Imielinski, C.M. Rice, A.C. Borczuk, C. Meydan, R.E. Schwartz, and C.E. Mason, *System-wide transcriptome damage and tissue identity loss in COVID-19 patients*. Cell Rep Med, 2022. **3**(2): p. 100522.

218. Sebastian, J., A. Thomas, C. Levine, R. Shrestha, S. Levy, H. Safi, S.R. Pentakota, P. Kumar, and D. Alland, *Origin and Dynamics of Mycobacterium tuberculosis Subpopulations That Predictably Generate Drug Tolerance and Resistance*. mBio, 2022. **13**(6): p. e0279522.
219. Arslan, S., F.J. Garcia, M. Guo, M.W. Kellinger, S. Kruglyak, J.A. LeVieux, A.H. Mah, H. Wang, J. Zhao, C. Zhou, A. Altomare, J. Bailey, M.B. Byrne, C. Chang, S.X. Chen, B. Cho, C.N. Dennler, V.T. Dien, D. Fuller, R. Kelley, O. Khandan, M.G. Klein, M. Kim, B.R. Lajoie, B. Lin, Y. Liu, T. Lopez, P.T. Mains, A.D. Price, S.R. Robertson, H. Taylor-Weiner, R. Tippiana, A.B. Tomaney, S. Zhang, M. Abtahi, M.R. Ambroso, R. Bajari, A.M. Bellizzi, C.B. Benitez, D.R. Berard, L. Berti, K.N. Blease, A.P. Blum, A.M. Boddicker, L. Bondar, C. Brown, C.A. Bui, J. Calleja-Aguirre, K. Cappa, J. Chan, V.W. Chang, K. Charov, X. Chen, R.M. Constandse, W. Damron, M. Dawood, N. DeBuono, J.D. Dimalanta, L. Edoli, K. Elango, N. Faustino, C. Feng, M. Ferrari, K. Frankie, A. Fries, A. Galloway, V. Gavril, G.J. Gemmen, J. Ghadiali, A. Ghorbani, L.A. Goddard, A.R. Guetter, G.L. Hendricks, J. Hentschel, D.J. Honigfort, Y.T. Hsieh, Y.H. Hwang Fu, S.K. Im, C. Jin, S. Kabu, D.E. Kincade, S. Levy, Y. Li, V.K. Liang, W.H. Light, J.B. Lipsher, T.L. Liu, G. Long, R. Ma, J.M. Mailloux, K.A. Mandla, A.R. Martinez, M. Mass, D.T. McKean, M. Meron, E.A. Miller, C.S. Moh, R.K. Moore, J. Moreno, J.M. Neysmith, C.S. Niman, J.M. Nunez, M.T. Ojeda, S.E. Ortiz, J. Owens, G. Piland, D.J. Proctor, J.B. Purba, M. Ray, D. Rong, V.M. Saade, S. Saha, G.S. Tomas, N. Scheidler, L.H. Sirajudeen, S. Snow, G. Stengel, R. Stinson, M.J. Stone, K.J. Sundseth, E. Thai, C.J. Thompson, M. Tjioe, C.L. Trejo, G. Trieger, D.N. Truong, B. Tse, B. Voiles, H. Vuong, J.C. Wong, C.T. Wu, H. Yu, Y. Yu, M. Yu, X. Zhang, D. Zhao, G. Zheng, M. He and M. Previte, *Sequencing by avidity enables high accuracy with low reagent consumption*. Nat Biotechnol, 2023.
220. Green, S.J., T. Torok, J.E. Allen, E. Elie-Fadrosh, S.A. Jackson, S.C. Jiang, S.S. Levine, S. Levy, L.M. Schriml, W.K. Thomas, J.M. Wood, and S.W. Tighe, *Metagenomic Methods for Addressing NASA's Planetary Protection Policy Requirements on Future Missions: A Workshop Report*. Astrobiology, 2023. **23**(8): p. 897-907.
221. Tierney, B.T., J. Kim, E.G. Overbey, K.A. Ryon, J. Foox, M. Sierra, C. Bhattacharya, N. Damle, D. Najjar, J. Park, S. Garcia Medina, N. Huerbi, C. Meydan, J. Wain Hershberg, J. Qiu, A. Kleinman, G. Al Ghalith, M. MacKay, E.E. Afshin, R. Dhir, J. Borg, C. Gatt, N. Brereton, B. Readhead, S. Beyaz, K.J. Venkateswaran, K. Blease, J. Moreno, A. Boddicker, J. Zhao, B. Lajoie, R.T. Scott, A. Altomare, S. Kruglyak, S. Levy, G. Church, and C.E. Mason, *Viral activation and ecological restructuring characterize a microbiome axis of spaceflight-associated immune activation*. Res Sq, 2023.

Exhibit B

MATERIALS AND DATA CONSIDERED

Literature

Acencio, Milena M. P., Evaldo Marchi, Lisete R. Teixeira, Bruna Rocha Silva, Juliana Sanchez Silva, Carlos Sergio Rocha Silva, Vanessa Adelia Alvarenga, Leila Antonangelo, Francisco Suso Vargas, and Vera Luiza Capelozzi. "Talc Particles and Pleural Mesothelium Interface Modulate Apoptosis and Inflammation." *Pathology* 46, no. S2 (2014): S76.

Acheson, E D, M J Gardner, E C Pippard, and L P Grime. "Mortality of Two Groups of Women Who Manufactured Gas Masks from Chrysotile and Crocidolite Asbestos: A 40-Year Follow-Up." *British Journal of Industrial Medicine* 39, no. 4 (November 1982): 344–48.

Adami, H.O., Hsieh, C.C., Lambe, M., Trichopoulos, D., Leon, D., Persson, I., Ekbom, A., and Janson, P.O. (1994). Parity, age at first childbirth, and risk of ovarian cancer. *Lancet* 344, 1250-1254.

Agic, A., Xu, H., Finas, D., Banz, C., Diedrich, K., and Hornung, D. (2006). Is endometriosis associated with systemic subclinical inflammation? *Gynecol Obstet Invest* 62, 139-147.

Akhtar, Mohd Javed, Maqsood Ahamed, M.A. Majeed Khan, Salman A. Alrokayan, Iqbal Ahmad, and Sudhir Kumar. "Cytotoxicity and Apoptosis Induction by Nanoscale Talc Particles from Two Different Geographical Regions in Human Lung Epithelial Cells." *Environmental Toxicology* 29 (2012): 394–406. <https://doi.org/10.1002/tox.21766>.

Akhtar, Mohd Javed, Sudhir Kumar, Ramesh Chandra Murthy, Mohd Ashquin, Mohd Imran Khan, Govil Patil, and Iqbal Ahmad. "The Primary Role of Iron-Mediated Lipid Peroxidation in the Differential Cytotoxicity Caused by Two Varieties of Talc Nanoparticles on A549 Cells and Lipid Peroxidation Inhibitory Effect Exerted by Ascorbic Acid." *Toxicology in Vitro: An International Journal Published in Association with BIBRA* 24, no. 4 (June 2010): 1139–47. <https://doi.org/10.1016/j.tiv.2010.03.002>.

Albu, Cristina-Crenguta. "The Genetics of Ovarian Cancer." *SMGroup*, (November 3, 2017).

Arellano-Orden, Elena, Auxiliadora Romero-Falcon, Jose Martin Juan, Manuel Ocana Jurado, Francisco Rodriguez-Panadero, and Ana Montes-Worboys. "Small Particle-Size Talc Is Associated with Poor Outcome and Increased Inflammation in Thoracoscopic Pleurodesis." *Respiration* 86 (2013): 201–9. <https://doi.org/10.1159/000342042>.

"ATSDR - Toxicological Profile: Asbestos." Dated September 2001. Accessed August 16, 2018. <https://www.atsdr.cdc.gov/toxprofiles/tp.asp?id=30&tid=4>.

Aust, A.E., Cook, P.M., and Dodson, R.F. Morphological and chemical mechanisms of elongated mineral particle toxicities. *J Toxicol and Environ Health, Part B*. (2011) 14:40-75.

Balkwill, F., and Mantovani, A. (2001). Inflammation and cancer: back to Virchow? *Lancet* 357, 539-545.

Banerjee, S., and Kaye, S.B. (2013). New strategies in the treatment of ovarian cancer: current clinical perspectives and future potential. *Clin Cancer Res* 19, 961-968.

Barnes, D.R., and Antoniou, A.C. (2012). Unravelling modifiers of breast and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: update on genetic modifiers. *J Intern Med* 271, 331-343.

Begg, Melissa D., and Dana March. "Cause and Association: Missing the Forest for the Trees." *American Journal of Public Health* 108, No. 5 (May 2018): 620–620.

Belotte, J., Fletcher, N.M., Saed, M.G., Abusamaan, M.S., Dyson, G, Diamond, M.P., Saed, G.M. (2015) A single nucleotide polymorphism in catalase is strongly associated with ovarian cancer survival. *PLoS ONE* 10(8): e0135739. Doi: 10.1371/journal.pone.0135739.

Belotte, Jimmy, Nicole M. Fletcher, Awoniyi O. Awonuga, Mitchell Alexis, Husam M. Abu-Soud, Ghassan M. Saed, Michael P. Diamond, and Mohammed G. Saed. "The Role of Oxidative Stress in the Development of Cisplatin Resistance in Epithelial Ovarian Cancer." *Reproductive Sciences* 21, no. 4 (2014): 503–8.

Berek, J.S., Crum, C., and Friedlander, M. (2012). Cancer of the ovary, fallopian tube, and peritoneum. *Int J Gynaecol Obstet* 119 Suppl 2, S118-129.

Berge, Wera, Kenneth Mundt, Hung Luu, and Paolo Boffetta. "Genital Use of Talc and Risk of Ovarian Cancer: A Meta-Analysis." *European Journal of Cancer Prevention*, July 2017.

Berry, G., M. L. Newhouse, and J. C. Wagner. "Mortality from All Cancers of Asbestos Factory Workers in East London 1933-80." *Occupational and Environmental Medicine* 57, no. 11 (November 2000): 782–85.

Bertolotti, Marinella, Daniela Ferrante, Dario Mirabelli, Mario Botta, Marinella Nonnato, Annalisa Todesco, Benedetto Terracini, and Corrado Magnani. "[Mortality in the cohort of the asbestos cement workers in the Eternit plant in Casale Monferrato (Italy)]." *Epidemiologia E Prevenzione* 32, no. 4–5 (October 2008): 218–28.

Blount, A.M. (1991). Amphibole content of cosmetic and pharmaceutical talcs. *Environ Health Perspect* 94, 225-230.

Blumenkrantz, M. J., N. Gallagher, R. A. Bashore, and H. Tenckhoff. "Retrograde Menstruation in Women Undergoing Chronic Peritoneal Dialysis." *Obstetrics and Gynecology* 57, no. 5 (May 1981): 667–70.

Bodmer, M., Becker, C., Meier, C., Jick, S.S., and Meier, C.R. (2011). Use of metformin and the risk of ovarian cancer: a case-control analysis. *Gynecol Oncol* 123, 200-204.

Bonovas, S., Filioussi, K., and Sitaras, N.M. (2005). Do nonsteroidal anti-inflammatory drugs affect the risk of developing ovarian cancer? A meta-analysis. *Br J Clin Pharmacol* 60, 194-203.

Booth, M, V Beral, and P Smith. "Risk Factors for Ovarian Cancer: A Case-Control Study." *British Journal of Cancer* 60, No. 4 (October 1989): 592-98.

Boorman, G. A., and J. C. Seely. "The Lack of an Ovarian Effect of Lifetime Talc Exposure in F344/N Rats and B6C3F1 Mice." *Regulatory Toxicology and Pharmacology: RTP* 21, no. 2 (April 1995): 242-43.

Bowtell, D.D. (2010). The genesis and evolution of high-grade serous ovarian cancer. *Nat Rev Cancer* 10, 803-808.

Buz'Zard, Amber R., and Benjamin H. S. Lau. "Pycnogenol® Reduces Talc-Induced Neoplastic Transformation in Human Ovarian Cell Cultures." *Phytotherapy Research* 21, No. 6 (June 2007): 579-86.

Camargo, M.C., Stayner, L.T., Straif, K., Reina, M., Al-Alem, U., Demers, P.A., and Landrigan, P.J. (2011). Occupational exposure to asbestos and ovarian cancer: a meta-analysis. *Environ Health Perspect* 119, 1211-1217.

Carr, C.J. "Talc: Consumer Uses and Health Perspectives" 21 (1995): 211-15.

Casagrande, J.T., Louie, E.W., Pike, M.C., Roy, S., Ross, R.K., and Henderson, B.E. (1979). "Incessant ovulation" and ovarian cancer. *Lancet* 2, 170-173.

Chan, J.K., Cheung, M.K., Husain, A., Teng, N.N., West, D., Whittemore, A.S., Berek, J.S., and Osann, K. (2006). Patterns and progress in ovarian cancer over 14 years. *Obstet Gynecol* 108, 521-528.

Chang, Stella, and Harvey A. Risch. "Perineal Talc Exposure and Risk of Ovarian Carcinoma." *Cancer* 79, No. 12 (June 15, 1997): 2396-2401.

Charbonneau, B., Goode, E.L., Kalli, K.R., Knutson, K.L., and Derycke, M.S. (2013). The immune system in the pathogenesis of ovarian cancer. *Crit Rev Immunol* 33, 137-164.

Chen Yong. "Risk Factors for Epithelial Ovarian Cancer in Beijing, China." *International Journal of Epidemiology*, 21, No. 1 (1992): 23-29.

Chen, L-M, et al. "Epithelial Carcinoma of the Ovary, Fallopian Tube, and Peritoneum: Epidemiology and Risk Factors - UpToDate," 2018.
https://www.uptodate.com/contents/epithelial-carcinoma-of-the-ovary-fallopian-tube-and-peritoneum-epidemiology-and-risk-factors?search=Epithelial%20carcinoma%20of%20the%20ovary,%20fallopian%20tube,%20and%20peritoneum:%20Epidemiology%20and%20risk%20factors&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1.

Committee on Practice Bulletins-Gynecology, Committee on Genetics, Society of Gynecologic

Oncology. “Practice Bulletin No 182: Hereditary Breast and Ovarian Cancer Syndrome.” *Obstetrics and Gynecology* 130, no. 3 (2017): e110–26.

Cook, L. S., M. L. Kamb, and N. S. Weiss. “Perineal Powder Exposure and the Risk of Ovarian Cancer.” *American Journal of Epidemiology* 145, No. 5 (March 1, 1997): 459–65.

Coussens, L.M., Tinkle, C.L., Hanahan, D., and Werb, Z. (2000). MMP-9 supplied by bone marrow-derived cells contributes to skin carcinogenesis. *Cell* 103, 481-490.

Coussens, L.M., and Werb, Z. (2002). Inflammation and cancer. *Nature* 420, 860-867.

Cralley, L. J., M. M. Key, D. H. Groth, W. S. Lainhart, and R. M. Ligo. “Fibrous and Mineral Content of Cosmetic Talcum Products.” *American Industrial Hygiene Association Journal* 29, no. 4 (August 1968): 350–54.

Cramer, Daniel. “Perineal Talc Exposure and Subsequent Epithelial Ovarian Cancer: A Case-Control Study.” *Obstetrics & Gynecology*, (1999).

Cramer, Daniel, et al. “Genital Talc Exposure and Risk of Ovarian Cancer.” *Int. J. Cancer*: 81 (1999): 351-356.

Cramer, D. W., W. R. Welch, R. E. Scully, and C. A. Wojciechowski. “Ovarian Cancer and Talc: A Case-Control Study.” *Cancer* 50, no. 2 (July 15, 1982): 372–76.

Cramer, Daniel W., Linda Titus-Ernstoff, John R. McKolanis, William R. Welch, Allison F. Vitonis, Ross S. Berkowitz, and Olivera J. Finn. “Conditions Associated with Antibodies Against the Tumor-Associated Antigen MUC1 and Their Relationship to Risk for Ovarian Cancer.” *Cancer Epidemiology Biomarkers & Prevention* 14, no. 5 (May 1, 2005): 1125–31.

Cramer, Daniel W., Allison F. Vitonis, Kathryn L. Terry, William R. Welch, and Linda J. Titus. “The Association Between Talc Use and Ovarian Cancer: A Retrospective Case–Control Study in Two US States.” *Epidemiology* 27, No. 3 (May 2016): 334–46.

Crusz, Shanthini M., and Frances R Balkwill. “Inflammation and Cancer: Advances and New Agents.” *Nature Reviews Clinical Oncology* 12 (October 2015): 584–96.

Dixon, Suzanne C., Christina M. Nagle, Nicolas Wentzensen, Britton Trabert, Alicia Beeghly-Fadiel, Joellen M. Schildkraut, Kirsten B. Moysich, et al. “Use of Common Analgesic Medications and Ovarian Cancer Survival: Results from a Pooled Analysis in the Ovarian Cancer Association Consortium.” *British Journal of Cancer* 116, no. 9 (April 25, 2017): 1223–28.

Dvorak, H.F. (1986). Tumors: wounds that do not heal. Similarities between tumor stroma generation and wound healing. *N Engl J Med* 315, 1650-1659.

Egli, G. E., and M. Newton. “The Transport of Carbon Particles in the Human Female

Reproductive Tract.” *Fertility and Sterility* 12 (April 1961): 151–55.

Eng, Kevin H., J. Brian Szender, John Lewis Etter, Jasmine Kaur, Samantha Poblete, Ruea-Yea Huang, Qianqian Zhu, et al. “Paternal Lineage Early Onset Hereditary Ovarian Cancers: A Familial Ovarian Cancer Registry Study.” *PLoS Genetics* 14, no. 2 (February 2018): e1007194.

Fasching, Peter A., Simon Gayther, Leigh Pearce, Joellen M. Schildkraut, Ellen Goode, Falk Thiel, Georgia Chenevix-Trench, et al. “Role of Genetic Polymorphisms and Ovarian Cancer Susceptibility.” *Molecular Oncology* 3, No. 2 (April 2009): 171–81.

FDA. “Ltr to Samuel S. Epstein, M.D., RE: Docket Numbers 94P-0420 and FDA-2008-P-0309-0001 /CP,” April 1, 2017.

Fernandes, José Veríssimo, Ricardo Ney Oliveira Cobucci, Carlos André Nunes Jatobá, Thales Allyrio Araújo de Medeiros Fernandes, Judson Welber Veríssimo de Azevedo, and Josélio Maria Galvão de Araújo. “The Role of the Mediators of Inflammation in Cancer Development.” *Pathology & Oncology Research* 21, no. 3 (July 2015): 527–34.

Ferrante, Daniela, Marinella Bertolotti, Annalisa Todesco, Dario Mirabelli, Benedetto Terracini, and Corrado Magnani. “Cancer Mortality and Incidence of Mesothelioma in a Cohort of Wives of Asbestos Workers in Casale Monferrato, Italy.” *Environmental Health Perspectives* 115, no. 10 (October 2007): 1401–5.

Fiume, Monice M., Ivan Boyer, Wilma F. Bergfeld, Donald V. Belsito, Ronald A. Hill, Curtis D. Klaassen, Daniel C. Liebler, et al. “Safety Assessment of Talc as Used in Cosmetics.” *International Journal of Toxicology* 34, no. 1 suppl (July 1, 2015): 66S-129S.

Fletcher, N.M., Belotte, J., Saed, M.G., Memaj, I., Diamond, M.P., Morris, R.T., Saed, G.M. Specific point mutations in key redox enzymes are associated with chemoresistance in epithelial ovarian cancer. *Free Radical Biology and Medicine* (2017) 102:122-132.

Fletcher, Nicole., Ira Memaj, Ghassan M. Saed. “Talcum Powder Enhances Oxidative Stress in Ovarian Cancer Cells.” *Reproductive Sciences*, 25 Suppl. 1 (March 2018): 214A-15A.

Fletcher, N.M., Saed, G.M. (2018). Talcum Powder enhances cancer antigen 125 levels in ovarian cancer cells and in normal ovarian epithelial cells. LB-044. Poster presented at: Society for Reproductive Investigation. 65th annual meeting. San Diego, CA.

Folkins, Ann K. “Chapter 24 - Assessing Pelvic Epithelial Cancer Risk and Intercepting Early Malignancy,” *Diagnostic Gynecologic and Obstetric Pathology (Third Edition)*, (2018): 844-864.

Franklin, R.A., and Li, M.O. (2016). Ontogeny of Tumor-associated Macrophages and Its Implication in Cancer Regulation. *Trends Cancer* 2, 20-34.

Freedman, Ralph S, Michael Deavers, Jinsong Liu, and Ena Wang. “Peritoneal Inflammation – A

Microenvironment for Epithelial Ovarian Cancer (EOC).” *Journal of Translational Medicine* 2, no. 23 (2004).

Friebel, Tara M., Susan M. Domchek, and Timothy R. Rebbeck. “Modifiers of Cancer Risk in BRCA1 and BRCA2 Mutation Carriers: Systematic Review and Meta-Analysis.” *Journal of the National Cancer Institute* 106, no. 6 (June 2014): dju091.

Galea, Sandro and Roger D. Vaughan. “Moving Beyond the Cause Constraint: A Public Health of Consequence.” *AJPH*, 108, No. 5 (May 2018): 602-603.

Gates, M. A., B. A. Rosner, J. L. Hecht, and S. S. Tworoger. “Risk Factors for Epithelial Ovarian Cancer by Histologic Subtype.” *American Journal of Epidemiology* 171, No. 1 (2010): 45–53.

Gates, M. A., S. S. Tworoger, K. L. Terry, L. Titus-Ernstoff, B. Rosner, I. De Vivo, D. W. Cramer, and S. E. Hankinson. “Talc Use, Variants of the GSTM1, GSTT1, and NAT2 Genes, and Risk of Epithelial Ovarian Cancer.” *Cancer Epidemiol Biomarkers Prev.* 17, No. 9 (September 2008): 2436–44.

Germani, D., S. Belli, C. Bruno, M. Grignoli, M. Nesti, R. Pirastu, and P. Comba. “Cohort Mortality Study of Women Compensated for Asbestosis in Italy.” *American Journal of Industrial Medicine* 36, no. 1 (July 1999): 129–34.

Gertig, Dorota M., David J. Hunter, Daniel W. Cramer, Graham A. Colditz, Frank E. Speizer, Walter C. Willett, and Susan E. Hankinson. “Prospective Study of Talc Use and Ovarian Cancer.” *J Natl Cancer Inst* 92, No. 3 (February 2, 2000): 249–52.

Ghio, Andrew J., Joleen M. Soukup, Lisa A. Dailey, Judy H. Richards, Jennifer L. Turi, Elizabeth N. Pavlisko, and Victor L. Roggli. “Disruption of Iron Homeostasis in Mesothelial Cells after Talc Pleurodesis.” *American Journal of Respiratory Cell and Molecular Biology* 46, no. 1 (January 1, 2012): 80–86.

Godard, Beatrice, William D. Foulkes, Diane Provencher, Jean-Sebastien Brunet, Patricia N. Tonin, Anne-Marie Mes-Masson, Steven A. Narod, and Parviz Ghadirian. “Risk Factors for Familial and Sporadic Ovarian Cancer among French Canadians: A Case-Control Study.” *Am J Obstet Gynecol* 179, No. 2 (August 1998): 403–10.

Gonzalez, Nicole L., Katie M. O’Brien, Aimee A. D’Aloisio, Dale P. Sandler, and Clarice R. Weinberg. “Douching, Talc Use, and Risk of Ovarian Cancer.” *Epidemiology* 27, No. 6 (November 2016): 797–802.

Gordon, Ronald E., Sean Fitzgerald, and James Millette. “Asbestos in Commercial Cosmetic Talcum Powder as a Cause of Mesothelioma in Women.” *International Journal of Occupational and Environmental Health* 20, no. 4 (October 2014): 318–32.

Green, Adèle, David Purdie, Christopher Bain, Victor Siskind, Peter Russell, Michael Quinn, and Bruce Ward. “Tubal Sterilisation, Hysterectomy and Decreased Risk of Ovarian Cancer.”

Int. J Cancer 71, No. 6 (June 11, 1997): 948–51.

Grivennikov, Sergei I., Florian R. Greten, and Michael Karin. “Immunity, Inflammation, and Cancer.” *Cell* 140, no. 6 (March 19, 2010): 883–99.

Gross, Alan J. and Paul H. Berg. “A Meta-Analytical Approach Examining the Potential Relationship Between Talc Exposure and Ovarian Cancer.” *Journal of Exposure Analysis and Environmental Epidemiology*, 5, No. 2 (1995): 181-195.

Halme, J., M. G. Hammond, J. F. Hulka, S. G. Raj, and L. M. Talbert. “Retrograde Menstruation in Healthy Women and in Patients with Endometriosis.” *Obstetrics and Gynecology* 64, no. 2 (August 1984): 151–54.

Hamilton, T.C., Fox, H., Buckley, C.H., Henderson, W.J., and Griffiths, K. Effects of talc on the rat ovary. *Br. J. Exp. Path.* (1984) 65: 101-106.

Harlow, Bernard L., and Noel S. Weiss. “A Case-Control Study of Borderline Ovarian Tumors: The Influence of Perineal Exposure to Talc.” *American Journal of Epidemiology* 130, No. 2 (August 1989): 390–94.

Harlow, Bernard L., Daniel W. Cramer, Debra A. Bell and William R. Welch. “Perineal Exposure to Talc and Ovarian Cancer Risk.” *Obstet Gynecol* 80, No. 1 (1992): 19-26.

Harper, A.K., Saed, G.M. Talc induces a pro-oxidant state in normal and ovarian cancer cells through gene point mutations in key redox enzymes. Accepted for presentation at SGO meeting 2019. [In Press].

Hartge, Patricia, Robert Hoover, Linda Leshner and Larry McGowan. “Talc and Ovarian Cancer.” *JAMA*, 250, No. 14 (1983): 1844.

Hein, M.J., Stayner, L.T., Lehman, E., Dement, J.M. Follow-up study of chrysotile textile workers: cohort mortality and exposure-response. *Occup Environ Med* (2007) 64:616-625.

Heller, D.S., Gordon, R.E., Westhoff, C., and Gerber, S. (1996). Asbestos exposure and ovarian fiber burden. *Am J Ind Med* 29, 435-439.

Henderson, W. J., T. C. Hamilton, M. S. Baylis, C. G. Pierrepont, and K. Griffiths. “The Demonstration of the Migration of Talc from the Vagina and Posterior Uterus to the Ovary in the Rat.” *Environmental Research* 40, no. 2 (August 1986): 247–50.

Henderson, W. J., C. A. Joslin, A. C. Turnbull, and K. Griffiths. “Talc and Carcinoma of the Ovary and Cervix.” *The Journal of Obstetrics and Gynaecology of the British Commonwealth* 78, no. 3 (March 1971): 266–72.

Hernán, Miguel A. “The C-Word: Scientific Euphemisms Do Not Improve Causal Inference From Observational Data.” *American Journal of Public Health* 108, No. 5 (May 2018): 616–19.

Hesterberg, T.W., Butterick, C.J., Oshimura, M., Brody, A.R., and Barrett, J.C. (1986). Role of phagocytosis in Syrian hamster cell transformation and cytogenetic effects induced by asbestos and short and long glass fibers. *Cancer Res* 46, 5795-5802.

Hill, Austin Bradford. "The Environment and Disease: Association or Causation?" *Proceedings of the Royal Society of Medicine* 58, no. 5 (May 1965): 295–300.

Hillegass, Jedd M., Arti Shukla, Maximilian B. MacPherson, Jeffrey P. Bond, Chad Steele, and Brooke T. Mossman. "Utilization of Gene Profiling and Proteomics to Determine Mineral Pathogenicity in a Human Mesothelial Cell Line (LP9/TERT-1)." *Journal of Toxicology and Environmental Health. Part A* 73, no. 5 (January 2010): 423–36.

Houghton, S. C., K. W. Reeves, S. E. Hankinson, L. Crawford, D. Lane, J. Wactawski-Wende, C. A. Thomson, J. K. Ockene, and S. R. Sturgeon. "Perineal Powder Use and Risk of Ovarian Cancer." *Journal of the National Cancer Institute* 106, No. 9 (September 10, 2014).

Huncharek, Michael, J.F. Geschwind and Bruce Kupelnick. "Perineal Application of Cosmetic Talc and Risk of Invasive Epithelial Ovarian Cancer: A Meta-analysis of 11,933 Subjects from Sixteen Observational Studies." *Anticancer Research*, 25 (2003): 1955-1960.

Huncharek, Michael, Joshua Muscat, Adedayo Onitilo, and Bruce Kupelnick. "Use of Cosmetic Talc on Contraceptive Diaphragms and Risk of Ovarian Cancer: A Meta-Analysis of Nine Observational Studies:" *European Journal of Cancer Prevention* 16, No. 5 (October 2007): 422–29.

Hunn, Jessica, and Gustavo C. Rodriguez. "Ovarian Cancer: Etiology, Risk Factors, and Epidemiology." *Clinical Obstetrics and Gynecology* 55, No. 1 (March 2012): 3–23.

Institute of Medicine (US) Committee on Asbestos: Selected Health Effects. *Asbestos: Selected Cancers*. The National Academies Collection: Reports Funded by National Institutes of Health. Washington (DC): National Academies Press (US), 2006.

International Agency for Research on Cancer (IARC). "IARC Monographs on the Evaluation of Carcinogenic Risks to Humans – IARC : Asbestos," 1977.

International Agency for Research on Cancer (IARC), "Cobalt in Hard Metals and Cobalt Sulfate, Gallium Arsenide, Indium Phosphide and Vanadium Pentoxide," IARC Monograph No. 86 (2006).

International Agency for Research on Cancer (IARC), "Overall Evaluations of Carcinogenicity: An Updating of IARC Monographs Volumes 1 to 42", IARC Supplement No. 7 (1987).

International Agency for Research on Cancer (IARC), “Silica and Some Silicates”, IARC Monograph No. 42 (1987).

International Agency for Research on Cancer (IARC), “Carbon Black, Titanium Dioxide, and Talc”, IARC Monograph No. 93 (2010).

International Agency for Research on Cancer (IARC), “Arsenic, Metals, Fibres, and Dusts”, IARC Monograph No. 100C (2012).

International Agency for Research on Cancer (IARC), “Mechanisms of Fibre Carcinogenesis”, IARC Scientific Publications, No. 140 (1996).

Irie, H., Banno, K., Yanokura, M., Iida, M., Adachi, M., Nakamura, K., Umene, K., Nogami, Y., Masuda, K., Kobayashi, Y., *et al.* (2016). Metformin: A candidate for the treatment of gynecological tumors based on drug repositioning. *Oncol Lett* 11, 1287-1293.

Iturralde, M., and P. F. Venter. “Hysterosalpingo-Radionuclide Scintigraphy (HERS).” *Seminars in Nuclear Medicine* 11, no. 4 (October 1981): 301–14.

Jaurand, M.C. Mechanisms of fiber-induced genotoxicity. *Environ Health Perspect* (1997) 105(Suppl 5):1073-1084.

Jayson, G.C., Kohn, E.C., Kitchener, H.C., and Ledermann, J.A. (2014). Ovarian cancer. *Lancet* 384, 1376-1388.

Jervis, Sarah, Honglin Song, Andrew Lee, Ed Dicks, Jonathan Tyrer, Patricia Harrington, Douglas F. Easton, Ian J. Jacobs, Paul P. D. Pharoah, and Antonis C. Antoniou. “Ovarian Cancer Familial Relative Risks by Tumour Subtypes and by Known Ovarian Cancer Genetic Susceptibility Variants.” *Journal of Medical Genetics* 51, no. 2 (February 2014): 108–13.

Jia, D., Nagaoka, Y., Katsumata, M., and Orsulic, S. (2018). Inflammation is a key contributor to ovarian cancer cell seeding. *Sci Rep* 8, 12394.

Jones, Richard E., and Kristin H. Lopez. “Human Reproductive Biology - 4th Edition Chapter 9 - Gamete Transport and Fertilization.” In *Human Reproductive Biology*, Third., 159–73. San Diego: Academic Press, 2006.

Jordan, S.J., Cushing-Haugen, K.L., Wicklund, K.G., Doherty, J.A., and Rossing, M.A. (2012). Breast-feeding and risk of epithelial ovarian cancer. *Cancer Causes Control* 23, 919-927.

Karageorgi, S., M. A. Gates, S. E. Hankinson, and I. De Vivo. “Perineal Use of Talcum Powder and Endometrial Cancer Risk.” *Cancer Epidemiology Biomarkers & Prevention* 19, No. 5 (May 1, 2010): 1269–75.

Kauff, Noah D., Nandita Mitra, Mark E. Robson, Karen E. Hurley, Shaokun Chuai, Deborah Goldfrank, Eve Wadsworth, et al. “Risk of Ovarian Cancer in BRCA1 and BRCA2

Mutation-Negative Hereditary Breast Cancer Families.” *Journal of the National Cancer Institute* 97, no. 18 (September 21, 2005): 1382–84.

Keskin, N., Teksen, Y.A., Ongun, E.G., Ozay, Y., Saygili, H. Does long-term talc exposure have a carcinogenic effect on the female genital system of rats? An experimental pilot study. *Arch Gynecol Obstet.* (2009). 280:925-931.

Kisielewski, R., Tolwinska, A., Mazurek, A., and Laudanski, P. (2013). Inflammation and ovarian cancer--current views. *Ginekol Pol* 84, 293-297.

Kissler, Stefan, Ernst Siebzehnuebl, Joachim Kohl, Anja Mueller, Nadja Hamscho, Regine Gaetje, Andre Ahr, Achim Rody, and Manfred Kaufmann. “Uterine Contractility and Directed Sperm Transport Assessed by Hysterosalpingoscintigraphy (HSSG) and Intrauterine Pressure (IUP) Measurement.” *Acta Obstetricia Et Gynecologica Scandinavica* 83, no. 4 (April 2004): 369–74.

Khan, Mohd Imran, AmoghA. Sahasrabuddhe, Govil Patil, Mohd Javed Akhtar, Mohd Ashquin, and Iqbal Ahmad. “Nano-Talc Stabilizes TNF- α m-RNA in Human Macrophages.” *Journal of Biomedical Nanotechnology* 7, no. 1 (January 1, 2011): 112–13.

Kiraly, Orsolya, Guanyu Gong, Werner Olipitz, Sureshkumar Muthupalani, and Bevin P. Engelward. “Inflammation-Induced Cell Proliferation Potentiates DNA Damage-Induced Mutations In Vivo.” *PLoS Genetics*, February 3, 2015.

Kunz, G., D. Beil, H. Deiniger, A. Einspanier, G. Mall, and G. Leyendecker. “The Uterine Peristaltic Pump. Normal and Impeded Sperm Transport within the Female Genital Tract.” *Advances in Experimental Medicine and Biology* 424 (1997): 267–77.

Kurta, M. L., K. B. Moysich, J. L. Weissfeld, A. O. Youk, C. H. Bunker, R. P. Edwards, F. Modugno, R. B. Ness, and B. Diergaarde. “Use of Fertility Drugs and Risk of Ovarian Cancer: Results from a U.S.-Based Case-Control Study.” *Cancer Epidemiol Biomarkers Prev.* 21, No. 8 (August 1, 2012): 1282–92.

Landen, Charles N., Michael J. Birrer, and Anil K. Sood. “Early Events in the Pathogenesis of Epithelial Ovarian Cancer.” *Journal of Clinical Oncology: Official Journal of the American Society of Clinical Oncology* 26, no. 6 (February 20, 2008): 995–1005.

Langseth, H., Johansen, B.V., Nesland, J.M., and Kjaerheim, K. (2007). Asbestos fibers in ovarian tissue from Norwegian pulp and paper workers. *Int J Gynecol Cancer* 17, 44-49.

Langseth, H, S E Hankinson, J Siemiatycki, and E Weiderpass. “Perineal Use of Talc and Risk of Ovarian Cancer.” *J Epidemiol Community Health* 62, No. 4 (April 1, 2008): 358–60.

Langseth, Hilde, and Kristina Kjaerheim. “Ovarian Cancer and Occupational Exposure among Pulp and Paper Employees in Norway.” *Scand J Work, Environ Health* 30, No. 5 (October 2004): 356–61.

Lee, Jennifer S., Esther M. John, Valerie McGuire, Anna Felberg, Kimberly L. Ostrow, Richard A. DiCioccio, Frederick P. Li, Alexander Miron, Dee W. West, and Alice S. Whittemore. "Breast and Ovarian Cancer in Relatives of Cancer Patients, with and without BRCA Mutations." *Cancer Epidemiology, Biomarkers & Prevention: A Publication of the American Association for Cancer Research, Cosponsored by the American Society of Preventive Oncology* 15, no. 2 (February 2006): 359–63.

Levanon, K., Crum, C., Drapkin, R. New insights into the pathogenesis of serous ovarian cancer and its clinical impact. *J Clin Oncol.* (2008). Nov 10;26(32):5284-93.

Li, D. (2011). Metformin as an antitumor agent in cancer prevention and treatment. *J Diabetes* 3, 320-327.

Lin, H.W., Tu, Y.Y., Lin, S.Y., Su, W.J., Lin, W.L., Lin, W.Z., Wu, S.C., and Lai, Y.L. (2011). Risk of ovarian cancer in women with pelvic inflammatory disease: a population-based study. *Lancet Oncol* 12, 900-904.

Liou, Geou-Yarh, and Peter Storz. "Reactive Oxygen Species in Cancer." *Free Radical Research* 44, no. 5 (May 2010): 476–96.

Liu, Y., and Cao, X. (2015). The origin and function of tumor-associated macrophages. *Cell Mol Immunol* 12, 1-4.

Lockey, J. E. "Nonasbestos Fibrous Minerals." *Clinics in Chest Medicine* 2, no. 2 (May 1981): 203–18.

Maccio, A., and Madeddu, C. (2012). Inflammation and ovarian cancer. *Cytokine* 58, 133-147.

Mabuchi, S., Kawase, C., Altomare, D.A., Morishige, K., Sawada, K., Hayashi, M., Tsujimoto, M., Yamoto, M., Klein-Szanto, A.J., Schilder, R.J., *et al.* (2009). mTOR is a promising therapeutic target both in cisplatin-sensitive and cisplatin-resistant clear cell carcinoma of the ovary. *Clin Cancer Res* 15, 5404-5413.

Magnani, C., D. Ferrante, F. Barone-Adesi, M. Bertolotti, A. Todesco, D. Mirabelli, and B. Terracini. "Cancer Risk after Cessation of Asbestos Exposure: A Cohort Study of Italian Asbestos Cement Workers." *Occupational and Environmental Medicine* 65, no. 3 (August 2007): 164–70.

Mahdavi, A., Pejovic, T., and Nezhat, F. (2006). Induction of ovulation and ovarian cancer: a critical review of the literature. *Fertil Steril* 85, 819-826.

Mäki-Nevala, Satu, Virinder Kaur Sarhadi, Aija Knuuttila, Ilari Scheinin, Pekka Ellonen, Sonja Lagström, Mikko Rönty, *et al.* "Driver Gene and Novel Mutations in Asbestos-Exposed Lung Adenocarcinoma and Malignant Mesothelioma Detected by Exome Sequencing." *Lung* 194, no. 1 (February 2016): 125–35.

Mallen, Adrienne R., Mary K. Townsend, and Shelley S. Tworoger. "Risk Factors for Ovarian Carcinoma." *Hematol Oncol Clin N Am*, September 2018.
<https://doi.org/10.1016/j.hoc.2018.07.002>.

Mantovani, A., Bottazzi, B., Colotta, F., Sozzani, S., and Ruco, L. (1992a). The origin and function of tumor-associated macrophages. *Immunol Today* 13, 265-270.

Mantovani, A., Bussolino, F., and Dejana, E. (1992b). Cytokine regulation of endothelial cell function. *FASEB J* 6, 2591-2599.

Mantovani, A., Bussolino, F., and Introna, M. (1997). Cytokine regulation of endothelial cell function: from molecular level to the bedside. *Immunol Today* 18, 231-240.

Melaiu, Ombretta, Federica Gemignani, and Stefano Landi. "The Genetic Susceptibility in the Development of Malignant Pleural Mesothelioma." *Journal of Thoracic Disease* 10, no. Suppl 2 (January 2018): S246–52.

Meng, Qingsong, Weixue Sun, John Jiang, Nicole M. Fletcher, Michael P. Diamond, and Ghassan M. Saed. "Identification of Common Mechanisms between Endometriosis and Ovarian Cancer." *Journal of Assisted Reproduction and Genetics* 28 (2011): 917–23.

Merritt, Melissa A., Adèle C. Green, Christina M. Nagle, Penelope M. Webb, and Australian Cancer Study (Ovarian Cancer) and Australian Ovarian Cancer Study Group. "Talcum Powder, Chronic Pelvic Inflammation and NSAIDs in Relation to Risk of Epithelial Ovarian Cancer." *Int. J. Cancer* 122, No. 1 (2008): 170–76.

Mills, P.K., Riordan, D.G., Cress, R.D., and Young, H.A. (2004). Perineal talc exposure and epithelial ovarian cancer risk in the Central Valley of California. *Int J Cancer* 112, 458-464.

Milne, R. L., and A. C. Antoniou. "Genetic Modifiers of Cancer Risk for BRCA1 and BRCA2 Mutation Carriers." *Annals of Oncology: Official Journal of the European Society for Medical Oncology* 22 Suppl 1 (January 2011): i11-17.

Milne, Roger L., and Antonis C. Antoniou. "Modifiers of Breast and Ovarian Cancer Risks for BRCA1 and BRCA2 Mutation Carriers." *Endocrine-Related Cancer* 23, no. 10 (2016): T69-84.
<https://doi.org/10.1530/ERC-16-0277>.

Modan, B., Hartge, P., Hirsh-Yechezkel, G., Chetrit, A., Lubin, F., Beller, U., Ben-Baruch, G., Fishman, A., Menczer, J., Struewing, J.P., *et al.* (2001). Parity, oral contraceptives, and the risk of ovarian cancer among carriers and noncarriers of a BRCA1 or BRCA2 mutation. *N Engl J Med* 345, 235-240.

Moon, Min Chaul, Jung Duck Park, Byung Soon Choi, So Young Park, Dong Won Kim, Yong Hyun Chung, Naomi Hisanaga, and Il Je Yu. "Risk Assessment of Baby Powder Exposure through Inhalation." *Toxicological Research* 27, no. 3 (September 2011): 137–41.

Moorman, P. G., R. T. Palmieri, L. Akushevich, A. Berchuck, and J. M. Schildkraut. "Ovarian

Cancer Risk Factors in African-American and White Women.” *Am J Epidemiol* 170, No. 5 (September 1, 2009): 598–606.

Mor, G., Yin, G., Chefetz, I., Yang, Y., and Alvero, A. (2011). Ovarian cancer stem cells and inflammation. *Cancer Biol Ther* 11, 708-713.

Mostafa, S. A., C. B. Barger, R. W. Flower, N. B. Rosenshein, T. H. Parmley, and J. D. Woodruff. “Foreign Body Granulomas in Normal Ovaries.” *Obstetrics and Gynecology* 66, no. 5 (November 1985): 701–2.

Muscat, J. E., and M. S. Huncharek. “Causation and Disease: Biomedical Science in Toxic Tort Litigation.” *Journal of Occupational Medicine: Official Publication of the Industrial Medical Association* 31, no. 12 (December 1989): 997–1002.

Nadler, Diana L., and Igor G. Zurbenko. “Estimating Cancer Latency Times Using a Weibull Model,” 2014, 8.

Narod, S.A., Risch, H., Moslehi, R., Dorum, A., Neuhausen, S., Olsson, H., Provencher, D., Radice, P., Evans, G., Bishop, S., *et al.* (1998). Oral contraceptives and the risk of hereditary ovarian cancer. Hereditary Ovarian Cancer Clinical Study Group. *N Engl J Med* 339, 424-428.

National Toxicology Program. Toxicology and carcinogenesis studies of talc (CAS No. 14807-96-6) in F344/N rats and B6C3F₁ mice (Inhalation studies); 1993. Report No. NTP TR 421; NIH Publication No. 93-3152.

Nelson, Heather H., and Karl T. Kelsey. “The Molecular Epidemiology of Asbestos and Tobacco in Lung Cancer.” *Oncogene* 21, no. 48 (October 21, 2002): 7284–88.

Ness, R. B., and C. Cottreau. “Possible Role of Ovarian Epithelial Inflammation in Ovarian Cancer.” *JNCI Journal of the National Cancer Institute* 91, no. 17 (September 1, 1999): 1459–67. <https://doi.org/10.1093/jnci/91.17.1459>.

Ness, Roberta B., Jeane Ann Grisso, Carrie Cottreau, Jennifer Klapper, Ron Vergona, James E. Wheeler, Mark Morgan, and James J. Schlesselman. “Factors Related to Inflammation of the Ovarian Epithelium and Risk of Ovarian Cancer.” *Epidemiology* 11, No. 2 (2000): 111–17.

Newhouse, M. L., G. Berry, J. C. Wagner, and M. E. Turok. “A Study of the Mortality of Female Asbestos Workers.” *British Journal of Industrial Medicine* 29, no. 2 (April 1972): 134–41.

NIOSH - CDC – Occupational Cancer – Carcinogen List, May 2012

“NIOSH 2011 Current Intelligence Bulletin No. 62,” 2011.

NIOSH. “Asbestos Fibers and Other Elongated Mineral Particles: State of the Science and Roadmap for Research (Revised Draft),” January 2009.

NIOSH. “Fiber Exposure during Use of Baby Powders, Report No. IWS-36-6,” July 1972..

“NTP Toxicology and Carcinogenesis Studies of Talc (CAS No. 14807-96-6)(NonAsbestiform) in F344/N.Rats and B6C3F1 Mice (Inhalation Studies),” 1993.

Nunes, S.C., and Serpa, J. (2018). Glutathione in Ovarian Cancer: A Double-Edged Sword. *Int J Mol Sci* 19.

Oberdörster, Günter, Eva Oberdörster, and Jan Oberdörster. “Nanotoxicology: An Emerging Discipline Evolving from Studies of Ultrafine Particles.” *Environmental Health Perspectives* 113, no. 7 (July 2005): 823–39.

Paoletti, L., Caiazza, S., Donelli, G., and Pocchiari, F. (1984). Evaluation by electron microscopy techniques of asbestos contamination in industrial, cosmetic, and pharmaceutical talcs. *Regul Toxicol Pharmacol* 4, 222-235.

Paluch-Shimon, S., Cardoso, F., Sessa, C., Balmana, J., Cardoso, M.J., Gilbert, F., Senkus, E., and Committee, E.G. (2016). Prevention and screening in BRCA mutation carriers and other breast/ovarian hereditary cancer syndromes: ESMO Clinical Practice Guidelines for cancer prevention and screening. *Ann Oncol* 27, v103-v110.

Paoletti, L., Caiazza, S., Donelli, G., and Pocchiari, F. (1984). Evaluation by electron microscopy techniques of asbestos contamination in industrial, cosmetic, and pharmaceutical talcs. *Regul Toxicol Pharmacol* 4, 222-235.

Pardoll, D.M. (2002). Spinning molecular immunology into successful immunotherapy. *Nat Rev Immunol* 2, 227-238.

Park, Hyo K., Joellen M. Schildkraut, Anthony J. Alberg, Elisa V. Bandera, Jill S. Barnholtz-Sloan, Melissa Bondy, Sydnee Crankshaw, et al. “Benign Gynecologic Conditions Are Associated with Ovarian Cancer Risk in African-American Women: A Case–Control Study.” *Cancer Causes & Control*, September 29, 2018.

Parmley, T. H., and J. D. Woodruff. “The Ovarian Mesothelioma.” *American Journal of Obstetrics and Gynecology* 120, no. 2 (September 15, 1974): 234–41

Peshkin, B., and et al. “Genetic Counseling and Testing for Hereditary Breast and Ovarian Cancer - UpToDate,” 2018.

https://www.uptodate.com/contents/genetic-counseling-and-testing-for-hereditary-breast-and-ovarian-cancer?search=Genetic%20counseling%20and%20testing%20for%20hereditary%20breast%20and%20ovarian%20cancer&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1.

Peshkin, B. and C. Isaacs. “Overview of Hereditary Breast and Ovarian Cancer Syndromes - UpToDate,” 2018.

https://www.uptodate.com/contents/overview-of-hereditary-breast-and-ovarian-cancer-syndromes?search=Overview%20of%20hereditary%20breast%20and%20ovarian%20cancer%20syndrome&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1.

Peshkin, B. and C. Isaacs. "Prevalence of BRCA1 and BRCA2 Mutations and Associated Cancer Risks - UpToDate," 2018.

https://www.uptodate.com/contents/prevalence-of-brca1-and-brca2-mutations-and-associated-cancer-risks?search=prevalence-of-brca1-and-brca2-mu%E2%80%A6search_result%26selectedTitle%3D1~73%26usage_type%3Ddefault%26display_rank%3D1&source=search_result&selectedTitle=2~150&usage_type=default&display_rank=2.

Pejovic, T., and Nezhat, F. (2011). Missing link: inflammation and ovarian cancer. *Lancet Oncol* 12, 833-834.

Penninkilampi, Ross and Guy D. Eslick. "Perineal Talc Use and Ovarian Cancer: A Systematic Review and Meta-Analysis." *Epidemiology* 29, No. 1 (January 2018): 41-49.

Phillips, J. C., P. J. Young, K. Hardy, and S. D. Gangolli. "Studies on the Absorption and Disposition of 3H-Labelled Talc in the Rat, Mouse, Guinea-Pig and Rabbit." *Food and Cosmetics Toxicology* 16, no. 2 (April 1978): 161-63.

Pinto, Mauricio, Carlos Balmaceda, Maria L. Bravo, Sumie Kato, Alejandra Villarroel, et al. "Patient Inflammatory Status and CD4+/CD8+ Intraepithelial Tumor Lymphocyte Infiltration are Predictors of Outcomes in High-Grade Serous Ovarian Cancer." *Gynecologic Oncology* (2018).

Pira, E, C Pelucchi, L Buffoni, A Palmas, M Turbiglio, E Negri, P G Piolatto, and C La Vecchia. "Cancer Mortality in a Cohort of Asbestos Textile Workers." *British Journal of Cancer* 92, no. 3 (February 2005): 580-86. <https://doi.org/10.1038/sj.bjc.6602240>.

Pira, Enrico, Canzio Romano, Francesco S. Violante, Andrea Farioli, Giovanna Spatari, Carlo La Vecchia, and Paolo Boffetta. "Updated Mortality Study of a Cohort of Asbestos Textile Workers." *Cancer Medicine* 5, no. 9 (2016): 2623-28.

Pukkala, Eero, Jan Ivar Martinsen, Elsebeth Lynge, Holmfridur Kolbrun Gunnarsdottir, Pär Sparén, Laufey Tryggvadottir, Elisabete Weiderpass, and Kristina Kjaerheim. "Occupation and Cancer - Follow-up of 15 Million People in Five Nordic Countries." *Acta Oncologica (Stockholm, Sweden)* 48, no. 5 (2009): 646-790.

Purdie, David, Adèle Green, Christopher Bain, Victor Siskind, Bruce Ward, Neville Hacker, Michael Quinn, Gordon Wright, Peter Russell, and Beatrice Susil. "Reproductive and Other Factors and Risk of Epithelial Ovarian Cancer: An Australian Case-Control Study." *International Journal of Cancer* 62, No. 6 (September 15, 1995): 678-84.

Radic, I, I Vucak, J Milosevic, A Marusic, S Vukicevic, and M Marusic. "Immunosuppression Induced by Talc Granulomatosis in the Rat." *Clinical and Experimental Immunology* 73, no. 2 (August 1988): 316-21.

Ramus, S.J., Vierkant, R.A., Johnatty, S.E., Pike, M.C., Van Den Berg, D.J., Wu, A.H., Pearce, C.L., Menon, U., Gentry-Maharaj, A., Gayther, S.A., *et al.* (2008). Consortium analysis of 7 candidate SNPs for ovarian cancer. *Int J Cancer* 123, 380-388.

Rebbeck, T.R., Mitra, N., Wan, F., Sinilnikova, O.M., Healey, S., McGuffog, L., Mazoyer, S., Chenevix-Trench, G., Easton, D.F., Antoniou, A.C., Nathanson, K.L., the CIMBA Consortium. Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA*. (2015). April 7; 313(13):1347-1361.

Reid, B.M., Permuth, J.B., Sellers, T.A. Epidemiology of ovarian cancer: a review. *Cancer Med Biol.* (February 2017) Vol 14, No 1.

Reid, A., J. Heyworth, N. de Klerk, and A. W. Musk. "The Mortality of Women Exposed Environmentally and Domestically to Blue Asbestos at Wittenoom, Western Australia." *Occupational and Environmental Medicine* 65, no. 11 (November 2008): 743-49.
<https://doi.org/10.1136/oem.2007.035782>.

Reid, A., N. de Klerk, and A. W. Musk. "Does Exposure to Asbestos Cause Ovarian Cancer? A Systematic Literature Review and Meta-Analysis." *Cancer Epidemiology Biomarkers & Prevention* 20, no. 7 (July 1, 2011): 1287-95.

Reid, Alison, Amanda Segal, Jane S. Heyworth, Nicholas H. de Klerk, and Arthur W. Musk. "Gynecologic and Breast Cancers in Women after Exposure to Blue Asbestos at Wittenoom." *Cancer Epidemiology, Biomarkers & Prevention: A Publication of the American Association for Cancer Research, Cosponsored by the American Society of Preventive Oncology* 18, no. 1 (January 2009): 140-47.

Reuter, Simone, Subash C. Gupta, Madan M. Chaturvedi, and Bharat B. Aggarwal. "Oxidative Stress, Inflammation, and Cancer: How Are They Linked?" *Free Radical Biology and Medicine* 49, no. 11 (December 1, 2010): 1603-16.

Ring, Kari L., Christine Garcia, Martha H. Thomas, and Susan C. Modesitt. "Current and Future Role of Genetic Screening in Gynecologic Malignancies." *American Journal of Obstetrics and Gynecology* 217, no. 5 (2017): 512-21.

Rohl, A. N. "Asbestos in Talc." *Environmental Health Perspectives* 9 (December 1974): 129-32.

Rohl, A. N., A. M. Langer, I. J. Selikoff, A. Tordini, R. Klimentidis, D. R. Bowes, and D. L. Skinner. "Consumer Talcums and Powders: Mineral and Chemical Characterization." *Journal of Toxicology and Environmental Health* 2, no. 2 (November 1976): 255-84.

Rosenblatt, Karin A., Moyses Szklo, and Neil B. Rosenshein. "Mineral Fiber Exposure and the Development of Ovarian Cancer." *Gynecologic Oncology* 45, No. 1 (April 1992): 20-25.

Rosenblatt, Karin A., Noel S. Weiss, Kara L. Cushing-Haugen, Kristine G. Wicklund, and Mary Anne Rossing. "Genital Powder Exposure and the Risk of Epithelial Ovarian Cancer." *Cancer*

Causes Control 22, No. 5 (May 2011): 737–42.

Rösler, J. A., H. J. Woitowitz, H. J. Lange, R. H. Woitowitz, K. Ulm, and K. Rödelberger. “Mortality Rates in a Female Cohort Following Asbestos Exposure in Germany.” *Journal of Occupational Medicine.: Official Publication of the Industrial Medical Association* 36, no. 8 (August 1994): 889–93.

Ross, M. “Geology, Asbestos, and Health.” *Environmental Health Perspectives* 9 (December 1974): 123–24.

Saed, Ghassan M., Rouba Ali-Fehmi, Zhong L. Jiang, Nicole M. Fletcher, Michael P. Diamond, Husam M. Abu-Soud, and Adnan R. Munkarah. “Myeloperoxidase Serves as a Redox Switch That Regulates Apoptosis in Epithelial Ovarian Cancer.” *Gynecologic Oncology* 116, no. 2 (February 2010): 276–81. <https://doi.org/10.1016/j.ygyno.2009.11.004>.

Saed, G.M., Diamond, M.P., and Fletcher, N.M. (2017). Updates of the role of oxidative stress in the pathogenesis of ovarian cancer. *Gynecol Oncol* 145, 595-602.

Saed, G.M., Morris, R.T., and Fletcher, N.M. (2018). New Insights into the Pathogenesis of Ovarian Cancer: Oxidative Stress. In *Ovarian Cancer-From Pathogenesis to Treatment* (IntechOpen).

Savant, Sudha S., Shruthi Sriramkumar and Heather M. O’Hagan. “The Role of Inflammation and Inflammatory Mediators in the Development, Progression, Metastasis, and Chemoresistance of Epithelial Ovarian Cancer.” *Cancers* 10, No. 251 (2018).

Schildkraut, J. M., S. E. Abbott, A. J. Alberg, E. V. Bandera, J. S. Barnholtz-Sloan, M. L. Bondy, M. L. Cote, et al. “Association between Body Powder Use and Ovarian Cancer: The African American Cancer Epidemiology Study (AACES).” *Cancer Epidemiol Biomarkers Prev.* 25, No. 10 (October 1, 2016): 1411–17.

Sellers, T.A., Huang, Y., Cunningham, J., Goode, E.L., Sutphen, R., Vierkant, R.A., Kelemen, L.E., Fredericksen, Z.S., Liebow, M., Pankratz, V.S., et al. (2008). Association of single nucleotide polymorphisms in glycosylation genes with risk of epithelial ovarian cancer. *Cancer Epidemiol Biomarkers Prev* 17, 397-404.

Shan, W., and Liu, J. (2009). Inflammation: a hidden path to breaking the spell of ovarian cancer. *Cell Cycle* 8, 3107-3111.

Shen, H., Fridley, B.L., Song, H., Lawrenson, K., Cunningham, J.M., Ramus, S.J., Cicek, M.S., Tyrer, J., Stram, D., Larson, M.C., et al. (2013). Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. *Nat Commun* 4, 1628.

Shukla, A., MacPherson, M.B., Hillegass, J., Ramos-Nino, M.E., Alexeeva, V., Vacek, P.M., Bond, J.P., Pass, H.I., Steele, C., Mossman, B.T. Alterations in gene expression in human mesothelial cells correlate with mineral pathogenicity. *Am J Respir Cell Mol Biol.* (2009). 41:114-123.

Shushan, Asher, Orn Pnlticl, and JoRe Iscovich. "Human Menopausal Gonadotropin and the Risk of Epithelial Ovarian Cancer." *Gynecol-Endocrin* 65, No. 1 (January 1996).

Siegel, R.L., Miller, K.D., and Jemal, A. (2015). Cancer statistics, 2015. *CA Cancer J Clin* 65, 5-29.

Sjösten, A. C. E., H. Ellis, and G. a. B. Edelstam. "Retrograde Migration of Glove Powder in the Human Female Genital Tract." *Human Reproduction* 19, no. 4 (April 1, 2004): 991–95.

Soong, Thing Rinda, Brooke E. Howitt, Alexander Miron, Neil S. Horowitz, Frank Campbell, Colleen M. Feltmate, Michael G. Muto, et al. "Evidence for Lineage Continuity between Early Serous Proliferations (ESPs) in the Fallopian Tube and Disseminated High-Grade Serous Carcinomas." *The Journal of Pathology*, July 25, 2018.

Starita, Lea M., Muhtadi M. Islam, Tapahsama Banerjee, Aleksandra I. Adamovich, Justin Gullingsrud, et al. "A Multiplex Homology-Directed DNA Repair Assay Reveals the Impact of More Than 1,000 BRCA1 Missense Substitution Variants on Protein Function." *American Journal of Human Genetics* 103, (October 4, 2018): 1-11.

Steiling, W., J. F. Almeida, H. Assaf Vandecasteele, S. Gilpin, T. Kawamoto, L. O'Keeffe, G. Pappa, K. Rettinger, H. Rothe, and A. M. Bowden. "Principles for the Safety Evaluation of Cosmetic Powders." *Toxicology Letters*, August 17, 2018.

Steiling, W., M. Bascompta, P. Carthew, G. Catalano, N. Corea, A. D'Haese, P. Jackson, et al. "Principle Considerations for the Risk Assessment of Sprayed Consumer Products." *Toxicology Letters* 227, no. 1 (May 16, 2014): 41–49.

Stewart, Louise M., Katrina Spilsbury, Susan Jordan, Colin Stewart, C. D'Arcy J. Holman, Aime Powell, Joanne Reekie, and Paul Cohen. "Risk of High-Grade Serous Ovarian Cancer Associated with Pelvic Inflammatory Disease, Parity and Breast Cancer." *Cancer Epidemiology* 55 (August 2018): 110–16.

Straif, Kurt, Lamia Benbrahim-Tallaa, Robert Baan, Yann Grosse, Béatrice Secretan, Fatiha El Ghissassi, Véronique Bouvard, et al. "A Review of Human Carcinogens—Part C: Metals, Arsenic, Dusts, and Fibres." *The Lancet Oncology* 10, No. 5 (May 2009): 453–54.

Terry, K. L., S. Karageorgi, Y. B. Shvetsov, M. A. Merritt, G. Lurie, P. J. Thompson, M. E. Carney, et al. "Genital Powder Use and Risk of Ovarian Cancer: A Pooled Analysis of 8,525 Cases and 9,859 Controls." *Cancer Prev Res* 6, No. 8 (August 2013): 811–21.

Titus-Ernstoff, L., Perez, K., Cramer, D.W., Harlow, B.L., Baron, J.A., and Greenberg, E.R. (2001). Menstrual and reproductive factors in relation to ovarian cancer risk. *Br J Cancer* 84, 714-721.

Torre, L.A., Bray, F., Siegel, R.L., Ferlay, J., Lortet-Tieulent, J., and Jemal, A. (2015). Global cancer statistics, 2012. *CA Cancer J Clin* 65, 87-108.

Trabert, B., R. B. Ness, W.-H. Lo-Ciganic, M. A. Murphy, E. L. Goode, E. M. Poole, L. A. Brinton, et al. "Aspirin, Nonaspirin Nonsteroidal Anti-Inflammatory Drug, and Acetaminophen Use and Risk of Invasive Epithelial Ovarian Cancer: A Pooled Analysis in the Ovarian Cancer Association Consortium." *J Natl Cancer Inst* 106, No. 2 (February 6, 2014).

Trabert, Britton, Elizabeth M Poole, Emily White, Kala Visvanathan, Hans-Olov Adami, Garnet L Anderson, Theodore M Brasky, et al. "Analgesic Use and Ovarian Cancer Risk: An Analysis in the Ovarian Cancer Cohort Consortium." *J Natl Cancer Inst*, 111, No. 2 (May 31, 2018).

Tzonou, Anastasia, Argy Polychronopoulou, Chung-cheng Hsieh, Apostolos Rebelakos, Anna Karakatsani and Dimitrios Trichopoulos. "Hair Dyes, Analgesics, Tranquilizers and Perineal Talc Application as Risk Factors for Ovarian Cancer," *Int. J. Cancer*, 55, (1993): 408-410.

U.S. Environmental Protection Agency (USEPA), "Arsenic, inorganic", in the Integrated Risk Information System database listing (1995). Available at the USEPA Website:
https://cfpub.epa.gov/ncea/iris/iris_documents/documents/subst/0278_summary

US EPA. "Health Assessment Document for Talc. | National Technical Reports Library - NTIS." -600/8-91/217, 1992.
<https://ntrl.ntis.gov/NTRL/dashboard/searchResults/titleDetail/PB92239524.xhtml>.

Vallyathan N.V. and J.E. Craighead, "Pulmonary pathology in workers exposed to nonasbestiform talc", *Human Pathology* 12(1):28-35 (1981).

Van Gosen, B. S., H.A. Lowers, S.J. Sutley, and C.A. Gent. "Using the Geologic Setting of Talc Deposits as an Indicator of Amphibole Asbestos Content." *Environmental Geology* 45, no. 7 (2004): 20.

Vasama-Neuvonen, K., E. Pukkala, H. Paakkulainen, P. Mutanen, E. Weiderpass, P. Boffetta, N. Shen, T. Kauppinen, H. Vainio, and T. Partanen. "Ovarian Cancer and Occupational Exposures in Finland." *American Journal of Industrial Medicine* 36, no. 1 (July 1999): 83–89.

Venkatesan, Priya. "Possible X Chromosome-Linked Transmission of Ovarian Cancer." *The Lancet. Oncology* 19, no. 4 (April 2018): e185.

Venter, P. F., and M. Iturralde. "Migration of a Particulate Radioactive Tracer from the Vagina to the Peritoneal Cavity and Ovaries." *South African Medical Journal = Suid-Afrikaanse Tydskrif Vir Geneeskunde* 55, no. 23 (June 2, 1979): 917–19.

Verdoodt, Freija, Christian Dehlendorff, Søren Friis, and Susanne K. Kjaer. "Non-Aspirin NSAID Use and Ovarian Cancer Mortality." *Gynecologic Oncology* 150, no. 2 (2018): 331–37.

Virta, RL. "The Phase Relationship of Talc and Amphiboles in a Fibrous Talc Sample." *IH*;

Report of Investigations, 1985.

Vitonis, A.F., Titus-Ernstoff, L., and Cramer, D.W. (2011). Assessing ovarian cancer risk when considering elective oophorectomy at the time of hysterectomy. *Obstet Gynecol* 117, 1042-1050.

Wang, N.S., Jaurand, M.C., Magne, L., Kheuang, L., Pinchon, M.C., and Bignon, J. (1987). The interactions between asbestos fibers and metaphase chromosomes of rat pleural mesothelial cells in culture. A scanning and transmission electron microscopic study. *Am J Pathol* 126, 343-349.

Wang, Q. (2016) "Cancer predisposition genes: molecular mechanisms and clinical impact on personalized cancer care: examples of Lynch and HBOC syndromes." *Acta Pharmacologica Sinica* 37:143-149.

Wehner, A.P. (1994). Biological effects of cosmetic talc. *Food Chem Toxicol* 32, 1173-1184.

Wehner, A. P., A. S. Hall, R. E. Weller, E. A. Lepel, and R. E. Schirmer. "Do Particles Translocate from the Vagina to the Oviducts and Beyond?" *Food and Chemical Toxicology: An International Journal Published for the British Industrial Biological Research Association* 23, no. 3 (March 1985): 367–72.

Wehner, A. P., R. E. Weller, and E. A. Lepel. "On Talc Translocation from the Vagina to the Oviducts and Beyond." *Food and Chemical Toxicology: An International Journal Published for the British Industrial Biological Research Association* 24, no. 4 (April 1986): 329–38.

Wendel, Jillian R. Hufgard, Xiyin Wang and Shannon M. Hawkins. "The Endometriotic Tumor Microenvironment in Ovarian Cancer." *Cancers* 10, No. 261 (2018).

Werner, I. "Presence of Asbestos in Talc Samples." *Atenschutzinform* 21, no. 5 (1982).

Whittemore, Alice S, Marion L Wu, Ralphs Paffenbarger, L Sarles, James B Kampert, and S'Tella Grosser. "Personal and Environmental Characteristics Related to Epithelial Ovarian Cancer," *Am J Epidemiol*, 128, No. 6 (1988) 1228-1240.

Wignall, B.K., and A.J. Fox. "Mortality of Female Gas Mask Assemblers." *British Journal of Industrial Medicine* 39, no. 1 (1982): 34–38.

Wong, C. "Perineal Talc Exposure and Subsequent Epithelial Ovarian Cancer: A Case-Control Study." *Obstetrics & Gynecology* 93, No. 3 (March 1999): 372–76.

Woodruff, J. D. "The Pathogenesis of Ovarian Neoplasia." *The Johns Hopkins Medical Journal* 144, no. 4 (April 1979): 117–20.

Wu, A.H., Pearce, C.L., Tseng, C.C., Templeman, C., and Pike, M.C. (2009). Markers of inflammation and risk of ovarian cancer in Los Angeles County. *Int J Cancer* 124, 1409-1415.

Wu, A. H., C. L. Pearce, C.-C. Tseng, and M. C. Pike. "African Americans and Hispanics Remain at Lower Risk of Ovarian Cancer Than Non-Hispanic Whites after Considering

Nongenetic Risk Factors and Oophorectomy Rates.” *Cancer Epidemiol Biomarkers Prev* 24, No. 7 (July 2015): 1094–1100.

Yegles, M., Saint-Etienne, L., Renier, A., Janson, X., and Jaurand, M.C. (1993). Induction of metaphase and anaphase/telophase abnormalities by asbestos fibers in rat pleural mesothelial cells in vitro. *Am J Respir Cell Mol Biol* 9, 186-191.

Yilmaz, Ercan, Mehmet Gul, Rauf Melekoglu, Isil Koleli. “Immunohistochemical Analysis of Nuclear Factor Kappa Beta Expression in Etiopathogenesis of Ovarian Tumors.” *Acta Cir Bras.* 33, No. 7 (2018): 641-650.

Zazenski, R., W. H. Ashton, D. Briggs, M. Chudkowski, J. W. Kelse, L. MacEachern, E. F. McCarthy, M. A. Nordhauser, M. T. Roddy, and N. M. Teetsel. “Talc: Occurrence, Characterization, and Consumer Applications.” *Regulatory Toxicology and Pharmacology: RTP* 21, no. 2 (April 1995): 218–29.

Zervomanoklakis, I, H.W. Ott, D Hadziomerovic, V. Mattle, B.E. Seeber, I. Virgolini, D. Heute, S. Kissler, G. Leyendecker, and L. Wildt. “Physiology of Upward Transport in the Human Female Genital Tract.” *Annals of New York Academy of Sciences* 1101, no. 1 (2007): 1–20.

AMA Analytical Services, Inc. – Certificate of Analysis – Job Name: Task 3 – Analysis of Official Samples; Job Number: CLIN 1 – Task 3 (Oct. 11, 2019).

Bayraktar, Soley et al. “Genotype-Phenotype Correlations by Ethnicity and Mutation Location in BRCA Mutation Carriers.” *Breast J.* vol. 21,3 (2015): 260-7

Centers for Disease Control and Prevention (CDC): Ovarian Cancer Statistics;
<https://www.cdc.gov/cancer/ovarian/statistics/index.htm>

Celebi, H. and Hilmi Bolat. “BRCA and Non-BRCA Variants Detected by Next Generation Sequencing in Patients with Hereditary Breast and/or Ovarian Cancer Syndrome.” *Acta Oncologica Turcica* 2022; 55: 77-84.

Coppa, Anna et al. “Novel and recurrent BRCA2 mutations in Italian breast/ovarian cancer families widen the ovarian cancer cluster region boundaries to exons 13 and 14.” *Breast Cancer Res Treat* vol. 148,3 (2014): 629-35.

Dakeng, S., et al. “BRCA1 and TP53 Mutations in Ovarian Cancer: Molecular Genetic Insights and Updated Situations in Thailand.” *Genomics and Genetics* 2018; 11(3): 26-34.

Emi, T. “Transcriptomic and Epigenomic Effects of Insoluble Particles on J774 Macrophages.” *Epigenetics* 2021; Vol. 16, No. 10, 1053-1070.

Fletcher, N., et al. (2019) “Molecular Basis Supporting the Association of Talcum Powder Use with Increased Risk of Ovarian Cancer.” *Reproductive Sciences*.

Harper, A., et al. "Talcum Powder Induces Malignant Transformation of Human Primary Normal Ovarian Epithelial Cells but not Human Primary Normal Peritoneal Fibroblasts." *Minerva Obstetrics and Gynecology* 2023 April;75(2):150-7.

Health Canada Finds Talcum Powder (news)(Dec 6th, 2018).

Health Canada Recall & Safety Alert – Talc – Potential Risk of Lung Effects and Ovarian Cancer (12/5/2018).

Health Canada, Draft Screening Assessment Talc.

Hurwitz LM, et al. "Association of Frequent Aspirin Use With Ovarian Cancer Risk According to Genetic Susceptibility." *JAMA Netw Open*. 2023;6(2):e230666.
doi:10.1001/jamanetworkopen.2023.0666

Mandarino, A., et al. "The Effect of Talc Particles on Phagocytes in Co-Culture with Ovarian Cancer Cells." *Environmental Research*, 2020; 180:108676.

O'Brien, K., et al. "Perineal Talc Use, Douching, and the Risk of Uterine Cancer." *Epidemiology* 2019;30: 845-852.

Rebbeck, T., et al. "Association of Type and Location of BRCA1 and BRCA2 Mutations with Risk of Breast and Ovarian Cancer." *JAMA* 313, no. 13 (April 7, 2015); 1347-61.

Rodriguez-Rojas, L., et al. "The Unique Spectrum of *MUYTH* Germline Mutations in Colombian Patients with Extracolonic Carcinomas." *The Application of Clinical Genetics* 2023;16 53-62.

Taher, M., et al. (2018) Systematic Review and Meta-Analysis of the Association between Perineal Use of Talc and Risk of Ovarian Cancer.

Taher, M., et al. (2018) Systematic Review and Meta-Analysis of the Association between Perineal Use of Talc and Risk of Ovarian Cancer – Supplemental Content.

Thompson, H.S., et al. (2002). "Psychosocial Predictors of BRCA Counseling and Testing Decisions Among Urban African-American Women." *Cancer Epidemiol Biomarkers Prev* 11(12): 1579-1585.

Toland, A.E., Brody, L.C. & the BIC Steering Committee. Lessons learned from two decades of *BRCA1* and *BRCA2* genetic testing: the evolution of data sharing and variant classification. *Genet Med* **21**, 1476–1480 (2019).

Weight of Evidence - General Principles and Current Applications at Health Canada (2011).

Woolen S., Lazar, A and Smith-Bindman, R. Association Between the Frequent Use of Perineal Talcum Powder Products and Ovarian Cancer: A Systematic Review and Meta-Analysis. *J. Gen Intern Med* 2022.

Woolen S., Lazar, A and Smith-Bindman, R. Association Between the Frequent Use of Perineal Talcum Powder Products and Ovarian Cancer: A Systematic Review and Meta-Analysis. Supplemental Content Online.

Wright, Jason. "What is New in Ovarian Cancer." *Obstet Gynecol* 2018; 132:1498-9.

Wu, S., et al. (2018) "Evaluating Intrinsic and Non-Intrinsic Cancer Risk Factors." *Nature Communications*.

Yang, X. et al. "Inherited Rare and Common Variants in PTCH1 and PTCH2 Contributing to the Predisposition to Reproductive Cancers." *Gene* 814 (2022) 146157.

Depositions/Trial Transcripts

Deposition of Alice M. Blount in Gail Lucille Ingham, et al. v. Johnson & Johnson, et al.

Depositions of John Hopkins (Aug 16 and 17, 2018; Oct 17, 2018; Nov 5, 2018); Hopkins Exhibit 28 and Exhibit D-1A

Deposition of Julie Pier (Sept. 12 and 13, 2018); Pier Exhibit 47

Deposition of Shawn Levy, PhD (January 11, 2019)

Expert Reports

Expert Report of Michael Crowley, PhD (Nov. 12, 2018)

Amended Expert Report of William E. Longo, PhD and Mark W. Rigler PhD (February 1, 2019)

Expert Report of William E. Longo, PhD and Mark W. Rigler PhD. Analysis of J&J Baby Powder & Valiant Shower to Shower Talc Products for Amphibole (Tremolite) Asbestos Expert Report. August 2, 2017.

Expert Report of William E. Longo, PhD and Mark W. Rigler PhD. TEM Analysis of Historical 1978 Johnson's Baby Powder Sample for Amphibole Asbestos. February 16, 2018.

Expert Report of William E. Longo, PhD, Mark W. Rigler, and William B. Egeland. "Below the Waist Application of Johnson & Johnson Baby Powder." Materials Analytical Service, LLC, September 2017.

Expert Report of Anne McTiernan, M.D., PhD (November 16, 2018)

Expert Report of Jack Siemiatycki, MSc, PhD (November 16, 2018)

Expert Report of Rebecca Smith-Bindman, MD (November 15, 2018)

Expert Report of Dr. Ghassan M. Saed (November 16, 2018)

Expert Report of Robert B. Cook, PhD (November 16, 2018)

Expert Report of Daniel L. Clarke-Pearson, MD (November 16, 2018)

Expert Report of Judith Wolf, MD (November 16, 2018)

Expert Report of Judith Zelikoff, PhD (November 16, 2018)

Expert Report of Arch Carson, MD, PhD (November 16, 2018)

Expert Report of Benjamin G. Neel, MD, PhD for General Causation Daubert Hearing (February 25, 2019)

Expert Report of Jeff Boyd, PhD for General Causation Daubert Hearing (February 25, 2019)

Documents Produced

JNJ 000018679-90

JNJTALC000864509-732

Medical Records

BondurantL-MDAMR-00872-00876

BondurantL-MDAMR-01167-01168

BondurantL-MDAMR-01177-01180

BondurantL-MDAMR-01262

BondurantL-TCCMR-01662-01668

BondurantL-TUMR-01245-01251

BondurantL-TUMR-01763-01773

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CONVERSE_HILARY_YALENEWHAVENHOSPITAL_01206-01222

ConverseH-FineE-00075-00081

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GALLARDO_A_WASHU_000167-000171

GALLARDO_A_WASHU_000207-000211

GALLARDO_ANNA_WASHU_MED_00324

JUDKINSC_DHMC_C_MDR000081-87
JUDKINSC_DHMC_C_MDR000414-418
JUDKINSC_DHMC_MDR000102-103
JUDKINSC_REC000003-9
JudkinsC-MGMR-00003-4

NEWSOME_CAPI_C_MDR000004-5
NEWSOME_REC000001-10
NEWSOMET_HCH_MDR000035-41

PRAUSAPL-000001-000006 (client path report)
PRAUSAPL-000010-000026
PRAUSAPL-MAYOC-002084-002085
RausaP-GenPathMR-00001-00013

Databases

<https://genome.ucsc.edu/cgi-bin/hgGateway> (Genome build "Dec 2013(GRCh38/hg38")
<https://www.ncbi.nlm.nih.gov/clinvar/>

Exhibit C

Shawn Levy, PhD

Medical Legal Testimony in last 4 years

Date: January 11, 2019

Johnson & Johnson Talcum Powder Products Marketing, Sales Practices and Product Liability
Litigation MDL No. 2738

Hourly Rate: \$500/hour